Quarantine Life

Hi - hope everyone is well and staying safe. Last March when I sent the last newsletter, I had no idea that in August we would be still in isolation and quarantine. I have heard that some states are doing better and are opening up – only to have virus outbreaks reoccur. Our families are being diligent, especially those with transplants. I am very hopeful that a vaccine will be safe and available for our kids by the end of the year!

I have an update to share regarding our family conference. We have decided not to host a virtual event this year, but plan to join HCU America Network support group for our next conference on June 26/27 in Bethesda, MD. More to come in our next newsletter.

OAA wanted to reach out to our families in the midst of this quarantine by offering N95 masks to families. We sent out 275 masks to 130 families in April/May. My helper, Melissa was kept busy with the packaging and mailing of the masks. We still have a few more – so if you need a couple, please send me an email.

Many of our young adults, who once were busy with sports, work or visiting friends are forced to stay indoors. OAA created a weekly “Young Adult Zoom Meet-Up” to engage this group of adults. I use the word “young” – but it’s open to anyone who is affected with an organic acid disorders age 16+. Send me an email if your ‘young’ adult would like to join us sometime!

Stay safe and well!!
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Stephanie Carleton and Josh McMahon
OAA Natural History Registry Coordinators
Financial Report

In the fall of each year, after filing the OAA annual 990 report with the IRS, the board of the OAA publishes the organization’s financial status. The statement shown here details years 2019 and 2018.

Since you were so generous, we have been able to make tremendous progress. Here are some of the highlights of the past year:

- Hosted a table at a Rare Disease Day Event at the University of Minnesota in February 2019
- Hosted a table at the SIMD Professional conference in Seattle, WA in April 2019
- Hosted a table at the Abbott Nutrition Metabolic Conference in Atlanta, GA in June 2019
- Hosted a table at the SIMD/NAMA (North American Metabolic Academy) training in the Twin Cities in September 2019
- Hosted at table at the Rare New England Conference in Portland, ME in October 2019
- Published and mailed three newsletters in 2019
- Traveled to Boston, MA to visit with pharmaceutical company, LogicBio and share information about OAA
- Attended the NORD IAMRARE Registry Leaders Community Meeting in Washington DC in October
- Awarded a $10,000 Grant to the NIH
- Awarded a $1,000 Grant to the Emory University to their Nutrition Research Program
- Awarded a $5,000 Grant to Aza Technology to assist in the production of a Portable Ammonia Device
- Collaborated with Rare Science, Inc. to send Rare Bears to 50 OAA children.

By all accounts, 2019 was a busy year for the OAA.
Since our last article update, we continue to study hereditary methylmalonic acidemia and cobalamin disorders in both the laboratory and the clinic. As many of you know, we have been actively studying one of the cobalamin disorders called combined methylmalonic acidemia and homocystinuria cblC type also known as cobalamin C deficiency or cblC. We reported a new zebrafish model of which was published in March 2020 in Human Molecular Genetics. This research project was the result of a collaboration between several groups at the National Human Genome Research Institute and the National Eye Institute and supported in part by The Michael Clapcich Fund for Retinal Research. If you would like to read the paper please go to this link: https://doi.org/10.1093/hmg/ddaa044

Individuals with cblC have a variety of clinical symptoms such as developmental delay and vision problems which are not well understood. The first patient with cblC was described in 1969 yet it took until 2006 for the gene MMACHC to be discovered by Dr. David Rosenblatt’s laboratory. Animal models help us study disease complications and new treatments and the gene discovery allowed Dr Rosenblatt to make mouse model of cblC. Unfortunately the mice with cblC died very early in their development when they were embryos.

Our group decided to take a different approach and create a zebrafish model of cblC. Zebrafish are interesting to use as an animal model for several reasons: 1) all organs develop in 4 days! 2) you can watch the embryos develop under the microscope 3) from one male and female you can generate hundreds of offspring (compared with <10 in mice) 4) zebrafish have about 70% of the same genes as humans including mmachc.

We used an approach called genome editing which allowed us to specifically cut the DNA of the cblC gene (mmachc) resulting in a deletion (some missing DNA letters) so that just like in patients, the mmachc gene does not work correctly (Figure 1). Then we bred fish that are carriers for the deletion so that 25% of the offspring had cblC and we observed their growth and development.

We learned a few things from studying our zebrafish with cblC: Without any treatments, the fish with cblC did not survive to adulthood. They appeared to develop normally during the first week but by four weeks of age they were extremely small, about 50% the size of their siblings (Figure 2). Their methylmalonic acid levels were extremely elevated by ~200 fold. We learned about the role of mmachc in the retina, the part of the eye that receives and interprets light. The zebrafish with cblC had small optic nerves and had less photoreceptors (the specific cells in the retina that respond to light). We also studied the gene expression in the eyes and found that a number of genes involved in blindness in humans were different in cblC zebrafish eyes vs their siblings. The genes we identified may help us learn more about what causes the eye disease in cblC. Finally, we treated the fish by adding to their tanks different types of B12 (cyanocobalamin, hydroxocobalamin, methylcobalamin), cystadane (betaine) and methionine. All of the treatments except cyanocobalamin showed a small improvement in the growth of the cblC fish, with methylcobalamin and methionine showing the most improvement overall.

It would be interesting to study these treatments in humans in combination with the standard therapies. Our zebrafish could be useful for studying new treatments in the future.

We thank all the participants in our clinical natural history study on MMA and cobalamin disorders over the past 16 years. We have made significant progress in our understanding of this group of disorders with your collective help. This progress has been directly inspired our participants, who motivate us to understand and hopefully someday to develop new therapies for cblC and other organic acidemias. We hope that everyone is staying well during these challenging days of the coronavirus pandemic.
My name is Connor. I have MMA CblA, an “inborn error of metabolism”, a “rare disease”. There is no cure; treatment consists of a protein restricted diet, oral medication, medical food supplements and intramuscular shots of B12. Living with MMA has always made me the odd man out. Whether it be ordering takeout and watching a football game or simply going out with friends for dinner, I know that I am different in a way that almost no one can understand.

I was born in 2001, before New York had newborn screening. When I was 3 days old, MMA almost killed me. After several weeks in the neonatal ICU and countless tests, my parents were finally given the diagnosis. They were told that I would not survive infancy, and if I did, I would be severely handicapped.

And so began a childhood that essentially meant just trying to live on to the next day. With little knowledge of how to handle this disease, my family lived on edge throughout my early years as an undercurrent of constant worry flowed through the house. I wondered why my friends went to the doctor when they didn’t feel well, but I went to the hospital. The common cold meant a trip to the emergency room. Vomiting or the flu could be a hospital admission.

My parent’s refusal to accept my grim prognosis took me to various specialists throughout the country. I eventually landed in a clinical study on MMA at the National Institutes of Health. It was here that I met Dr. Manoli, Dr. Venditti, Jennifer Sloan, and the rest of their team. They have been an invaluable source of information and support to me and my parents over the years.

As a child, I followed a vegetarian diet, before being a vegetarian was en vogue. I became adept at reading food labels and calculating the grams of protein. My diet is a balancing act to consume the exact amount of protein; not too much, and not too little as either scenario results in toxic buildup of acid. Growing up as a vegetarian was difficult since it was not as common of a diet as it is today. When other kids were eating hot dogs and hamburgers at pool parties, I was eating potato salad sandwiches; when my friends were enjoying chicken wings while watching a hockey game, I was eating the celery sticks that came along with them. My unusual eating habits became such an anomaly to my peers that I assumed the role of “the boy with the meat allergy” instead of having to explain the complicated nature of my disorder.

While it may seem that my dietary restrictions make my life less enjoyable, this could not be farther from the truth. I have never known any other way of life than the one I live now, so, in my eyes, it was normal. Once my diet and medication routine were established and it was apparent that I was metabolically stable, my life became that of any ordinary boy. I was determined to not let MMA get in the way of what I wanted to do. I took the most rigorous curriculum that my school offered, and worked hard to maintain my grades. I play on my high school varsity golf and hockey teams, and play alto saxophone in the band. I also belong to numerous school clubs and work during the summers as a caddy. I enjoy hanging out with my friends, playing cards, street hockey, video games, or just watching sports on TV.

Although a burden, dealing with my disease has taught me discipline, sacrifice, and empathy. It has exposed me to people I otherwise would not have encountered and gives me a perspective on life that has helped me to make the most out of my life, acutely aware that it is a gift some people are not able to enjoy. My parents have always told me to celebrate other people’s differences because that is what makes individuals unique. Being the odd man out has shown me how true this really is.

I graduated from high school in June and will be attending University of Notre Dame. I am excited to start this new chapter of my life. I was accepted into the Mendoza School of Business where I will pursue a degree in finance. I would be happy to speak to anyone about my experiences living with MMA. Feel free to contact me at cflynn101401@gmail.com.
Living with a rare disease can cause ongoing emotional distress. So much about a rare disease and its progression remains uncertain; treatments and dosing are often experimental. And, it can be tough finding someone knowledgeable enough about the disease to provide reliable, specialized care.

April Murphy, 46, has a rare genetic disorder called methylmalonic acidemia (MMA), a life-threatening metabolic condition that causes severe episodes of illness and long-term complications. Murphy currently is in end-stage renal failure, awaiting a kidney transplant. Despite her condition, she considers herself fortunate.

She visited NIH for the first time in January, when she met NHGRI senior investigator and MMA expert Dr. Charles Venditti, and reconnected with NICHD director Dr. Diana Bianchi, who had counseled and treated her and her mother for an unrelated genetic condition two decades earlier.

“As somebody who works with doctors daily,” said Murphy, a surgical assistant in Williamsburg, Va., “what NIH doctors do is not a job to them. It’s so much more. They put 110 percent of themselves into what they do to take care of people, and you don’t find that everywhere.”

First Fetal Patient

Murphy’s luck began before she was born. Growing up in the Boston area, she auspiciously came under the care of Dr. Mary Ampola, a biochemical geneticist at Tufts-New England Medical Center, who first treated Murphy in her mother’s womb.

Incredibly, just a few years before Murphy was born, a researcher determined that some patients with MMA responded to large amounts of vitamin B12 (cobalamin). That discovery inspired Ampola to try a revolutionary treatment after amniocentesis confirmed that Murphy had inherited the vitamin-responsive type of MMA.

In 1973, with support from NIH and March of Dimes grants, Murphy’s mother received oral, then intramuscular injections of cobalamin for the last 2 months of pregnancy; the vitamin entered the placenta to treat baby April. The groundbreaking case was reported in 1975 in the New England Journal of Medicine.

“At that time, MMA patients didn’t get diagnosed until symptoms appeared in infancy or later in childhood. In Murphy’s case, a family tragedy saved her life. Her older sister died at age 3 months. Posthumous testing revealed that her sibling had MMA. While the condition is now routinely tested as part of newborn screening, it’s still unusual to make a prenatal diagnosis.

“The cobalamin-A form of the disorder, what April has, within the spectrum of MMA is considered milder,” said Venditti, “but it’s
still extremely dangerous...With this type, there’s a partial response to B12, but if a patient doesn’t get enough vitamin, he or she can go into a coma or have other serious complications.”

Lucky in Life
Growing up, Murphy had a normal childhood. She logged her diet daily, knew her limitations and took full responsibility for managing her condition, all under the supportive watch of Ampola. Murphy did have some scares, and was hospitalized a few times for metabolic acidosis, but quickly recovered.

She has taken hydroxocobalamin most of her life. Terrified of needles, she’d squirt the liquid vitamin into ginger ale. Drinking the elixir daily maintained her methylmalonic acid levels.

After high school, Murphy gave birth to a healthy daughter, and now has two grandchildren. Her daughter, as well as her two younger sisters, are MMA carriers but none of them have the recessive disorder.

As an adult looking back, Murphy is grateful for the meticulous care she received, and the experimental prenatal treatment that contributed to her better outcome at birth and beyond.

“Doctors have told me, after all my testing, that there could have been neurocognitive damage, possibly physical issues,” said Murphy, “so it really made a difference that they [gave my mother the B12] in utero versus waiting until I was born.”

A Genetic Scare
Twenty years ago, Murphy and her mother met with Bianchi, who was launching a genetic research project at Tufts-New England Medical Center on cancer in families. Murphy’s mother had survived breast and ovarian cancer and today remains cancer-free. Bianchi would counsel and treat them after they both tested positive for the BRCA gene.

“I come from a long line of strong women,” said Murphy, undeterred by the diagnosis. “It was great to find out so early on so that I could make decisions for my future before anything happened.”

Ten years ago, a preventative oophorectomy sent Murphy into early menopause. The accompanying extreme fatigue lasted an unusually long time. It turned out, the fatigue was caused by dangerously high acid levels, which would take years to stabilize. A nephrologist later diagnosed her with end-stage renal failure.

Toward New MMA Treatments
NHGRI’s Venditti has devoted much of his career to studying MMA. He began seeing patients in 1999 while a genetics fellow at Children’s Hospital of Philadelphia.

Doctors tried to discourage him from working on MMA, arguing that it wasn’t treatable, but, encouraged by many parents, he was determined to find better therapies.
“Now we have a lot more options for the patients and I really think it won’t be long until we have gene therapy,” said Venditti. “We’re going to get this to work.”

He and his colleagues at NHGRI have teamed up with several biotech companies and are developing promising new genomic therapies for MMA. He also oversees an MMA natural history study in the Clinical Center that has enrolled more than 200 patients; Murphy is the newest member of the cohort.

“One needs to understand the natural history of MMA before a new treatment is tested,” said Venditti, “and answer the questions of what the effects will be downstream on the patient.” The NHGRI natural history study helps track metabolic parameters, nutrition patterns, bone phenotypes, symptoms over time, genetic effects and, in concert with lab work, has identified new biomarkers for treatment.

“I counseled April more than 20 years ago,” said Bianchi, “but to see her again, it’s such a privilege because you get that long-term perspective. It’s an amazing story because it’s a rare but treatable genetic disorder, and there have been a lot of advances that really translate to patient care.”

Looking Ahead

During Murphy’s recent NIH visit, Venditti and his NHGRI team members Dr. Irini Manoli and Dr. Jennifer Sloan reassured her that renal failure was a consequence of MMA and that there was nothing she could have done to stop the progression. She also came away comforted that NIH doctors and resources are just a few hours from home.

Murphy is scheduled for a kidney transplant this spring. Neither sister was a match, so she and her wife are participating in a kidney swap program. Murphy is otherwise deemed in good health and optimistic about the future.

“In Massachusetts, I grew up my whole life having Dr. Ampola as my hero, always there to take care of me,” said Murphy. “When I moved to Virginia, there was an emotional loss. It was hard to find anyone to take care of me here who knew anything about MMA.”

She worried about where she’d turn if her condition changed or if she had any sort of emergency. “Daily life here, medically, has been a scary thing. But…now I have Dr. Bianchi and Drs. Venditti and Manoli and the MMA team in Bethesda. If anything should go wrong, I have people I can count on to take care of me.”

In tears, Murphy added, “It really makes the biggest difference in my life. You guys truly are the superheroes of medicine.”

LETTER TO THE EDITOR:

Update from our friends in Italy

O ne month is passed quickly and our virtual walking was a great success. We still leave the page in order to get some more donations from friends during the rest of summer. We raised about 8000 euros coming from more than 600 virtual tickets. Moreover we also get a sponsorship from Nutricia Italy that we are still waiting for the exact amount but it should probably double the total amount.

It is now 13 years that we organize events and last month during the annual associates’ meeting we informed all our friends that in these years we reached about 700,000 euro of donations to the metabolic lab and ward of the Pediatric Hospital Bambino Gesù in Rome and we are very proud of this.

As for the research currently going on in the lab, the ward primary told us they are using the new orbitrap mass spectrometer and working with models for propionic and methylmalonic acidemias but we are not allowed to share names of private companies that asked for their collaboration.
Dear MMA and PA Community,

We are excited to inform you that the US Food and Drug Administration has cleared HemoShear to proceed with a clinical trial of our investigational drug HST5040 for the treatment of MMA and PA.

HST5040 is an oral therapy developed by HemoShear that will be tested to correct metabolic abnormalities associated with MMA and PA. HST5040 has the potential to be active throughout the body, including the brain, heart and muscles. The drug can be taken conveniently at home as a daily liquid formulation by mouth or through a gastric feeding tube.

Our clinical study, HERO (HElp Reduce Organic Acids), is designed to enroll at least 12 patients with MMA and PA. The study, which will start with patients age 12 and over and then expand to age 2 and older, will be conducted at select leading children’s hospitals in the United States.

We are in the process of contracting with study sites and plan to be ready to begin later this year. Our first priority is ensuring that all families are safe, so the timing of the first patient enrolled may be impacted by the COVID-19 pandemic.

You are a Hero!

Making medical progress to improve the quality of life for MMA and PA patients is going to be a collaborative process between industry, clinical researchers and families. We will be in touch with the community when the study is ready to begin enrolling participants. In the meantime, we invite you to visit our website and sign up if you want to receive updates from us.

We are humbled and excited to see if our drug can make a positive difference in patients’ lives.

On behalf of the entire HemoShear team, thank you for your interest and support.

Sincerely,

Brian Wamhoff, PhD
Co-Founder and Head of Innovation
HemoShear Therapeutics Receives FDA Fast Track and Rare Pediatric Disease Designations for HST5040 to Treat Methylmalonic Acidemia and Propionic Acidemia

CHARLOTTESVILLE, Va., July 28, 2020 /PRNewswire/ -- HemoShear Therapeutics a clinical stage company developing treatments for rare metabolic disorders, has received Fast Track and Rare Pediatric Disease designations from the U.S. Food and Drug Administration (FDA) for HST5040, a once-daily oral small molecule drug being developed to treat methylmalonic acidemia (MMA) and propionic acidemia (PA).

In June, HemoShear received clearance from the FDA for its Investigational New Drug application to conduct a phase 2 clinical study of HST5040 in patients with MMA and PA. MMA and PA are rare genetic disorders caused by the deficiency of certain enzymes required to metabolize amino acids. The diseases result in the rapid buildup of life-threatening metabolites that can lead to severe organ damage, seizures, developmental deficits, and premature death. HemoShear’s phase 2 clinical study, HERO (HElp Reduce Organic Acids), is designed to enroll at least 12 patients with MMA and PA at select children’s hospitals in the United States.

"It is very encouraging that the FDA has recognized the urgent need for developing therapies like HST5040 to improve the lives of patients with MMA and PA," said Nicola Longo, M.D., Ph.D., Chief of the Division of Medical Genetics at the University of Utah and Professor of Pediatrics at Primary Children’s Medical Center. "Clinicians on the front lines of treating these devastating diseases have very limited options to help their patients."

Fast Track designation is intended to accelerate the development and review of therapies to treat serious conditions with unmet medical need. This award will enable HemoShear to have more frequent interactions with the FDA, as well as eligibility for accelerated approval, priority review, and rolling submission of a new drug application. Rare Pediatric Disease designation is awarded by the FDA for therapies being developed to treat serious or life-threatening diseases that primarily affect children ages 18 years or younger and fewer than 200,000 people in the U.S. Through this award, HemoShear may receive a voucher upon commercial approval of HST5040 that provides for priority review of a subsequent new drug application.

"Knowing that there are no effective treatments currently available for patients, we are working hard to initiate our HERO clinical trial," said Jim Powers, Chairman and CEO of HemoShear. "We will be working closely with the MMA and PA communities to enroll patients into our study of this novel and convenient oral therapy."

About HST5040
HST5040 is an investigational oral small molecule therapy developed by HemoShear to address metabolic abnormalities associated with MMA and PA. Because HST5040 is a small molecule, it has the ability to distribute to multiple affected tissues and thus has the potential to be active throughout the body, including the brain, heart and muscles. HST5040 is designed for convenient daily administration at home as a liquid formulation taken either orally or through a gastric feeding tube.
EDUCATION CORNER

TEMPLE makes learning EASIER

MMA/PA TEMPLE booklet now available for download!

TEMPLE (standing for Tools Enabling Metabolic Parents Learning) is a leading education series for inborn errors of metabolism, including Methylmalonic/Propionic Acidemia (MMA/PA), Glutaric Aciduria Type 1 (GA-1), and more. These booklets and videos explain the condition and its management in easy-to-understand language and pictures. They are ideal for educating new parents and families after a positive newborn screening. TEMPLE booklets and videos can also be used to teach grandparents, relatives, and friends about the condition. TEMPLE was created by Nutricia.

Exciting news from Nutricia is that they released a new TEMPLE booklet for MMA/PA! It can be downloaded at nutricialearningcenter.com/globalassets/pdfs/metabolics/temple_mma-pa_en_apr2020-v2.pdf, for free.

Share TEMPLE with relatives and friends to help them better understand the basics of your child’s or loved one’s condition. You can find all TEMPLE booklets on Nutricia’s website and TEMPLE videos on Nutricia’s YouTube channel.

We would also like to acknowledge a few graduations! Congratulations to these young men! Jeffrey, Johnny, Trent and Connor graduated from high school this fall.

Congratulations!
Hi all OAA members,

Every couple years I like to give an update on my life. I like to share my positive experience, thoughts, and success stories to encourage all of you, who have MMA or any genetic metabolic condition for that fact. Everyone can make it, I truly believe prayers and faith go a long way.

In February 2019, God blessed me with a second bundle of joy; I have a second daughter now. She is cheerful, happy, and loves to play. I am currently working at Burger King on my way training as a manager. I am planning to go back to school for medical billing and coding online, so that I can find a better job in the near future. I have to tell everyone out there honestly, please follow doctor’s orders as far as medications, they will really take a toll on your body and health if you choose to discontinue them. We are all special. I hope everyone stays strong through this coronavirus pandemic.

Stay strong. Please feel free to contact me by email, do not hesitate, chefcarlinssia94@gmail.com any questions or just feel like talking email is the best way to reach me.

Enjoy the summer!

CARLINSSIA
Waste no day to achieve the impossible! When Pennsylvania started testing newborns for metabolic diseases, Adam Settle was one of the first to be diagnosed with Cobalamin-C Deficiency. Legally blind and mentally delayed, Adam never let his limitations hold him back from accomplishing his dreams. His love for people and his desire to help others comes through the pages of this book. Whatever has been holding you back, this book will help you charge through the obstacles you face to achieve the impossible. Adam’s story will inspire, encourage and give you hope.

Interview with Adam Settle

**How did the idea of writing No Day Wasted get started?**
The idea came from encouragement from others to tell my story. After the third person within a week, I decided it was time.

**Can you tell us about the process of having No Day Wasted written?**
I was at an Engage conference with 99 Balloons in Arkansas with a friend Ryan. He said to me “Adam, you don’t waste a day!” And the next thing I knew we decided that should be the name of the book!

**What was the hardest part of having the book written?**
Waiting and deciding how to publish the book!

**You have lived a very full and exciting life, and you aren’t that old - how did you decide what parts to have written about and what parts to hold back?**
The author decided we would start at the beginning of life, which of curse was a rocky one! All the way to graduation from high school and all the adventures in between!

**We encourage patients to share their stories, what would you say to patients and caregivers who might be a little bit hesitant to share their or their child’s story?**
When you share your life story, others know they are not alone. We are all loved and uniquely made. We all need encouragement and hope. I hope my book touches hearts and changes attitudes!

**Everyone has a story to share!**

You can buy a copy of Adam’s book at Amazon: [amazon.com/No-Day-Wasted-Settle-Story](amazon.com/No-Day-Wasted-Settle-Story).
WHAT IS NAVIGATE NEWBORN SCREENING?

Navigate Newborn Screening is a free, learning opportunity that gives families information on one of the most common tests newborns get - newborn screening. The module can help families just learning about screening as well as those looking to be leaders in this system.

In this module, you will learn about:

- The newborn screening process
- Newborn screening results
- Types of conditions detected
- Questions to ask your healthcare provider
- How to tell your newborn screening story
- Additional newborn screening resources

WHY LEARN ABOUT NEWBORN SCREENING?

- Newborn screening is a state-run public health service that ensures all babies are screened for certain conditions that can cause serious health problems.
- Newborn screening usually happens when your baby is between 24 and 48 hours.
- In the U.S, all states require newborn screening, but not every state screens for the same conditions.
- Only 1 in 3 people can correctly identify the definition of newborn screening.

BENEFITS OF PARTICIPATING

- Learn about the most common screening test
- Gain leadership and advocacy skills
- Options to attend national conferences or meetings

Sign up today at EXPECTINGHEALTH.MYABSORB.COM

Learn about the most common test newborns get - #newbornscreening!

Newborn screening is a state-run public health service that ensures all babies are screened for certain conditions that can cause serious health problems. Expecting Health is excited to announce the launch of Navigate Newborn Screening, a brand new educational module for families. Navigate Newborn Screening is free to access and shares important information about what newborn screening means for babies and their families. Sign up at expectinghealth.myabsorb.com?KeyName=NavigateNBBS_OAA

Interested but have more questions? Contact Annie Evans at aevans@geneticalliance.org
ADULTS WITH RARE DISORDERS SUPPORT STUDY: COVID-19 SURVEY

What is the study about?
The Adults with Rare Disorders Support (AWaRDS) project is the largest study aiming to improve quality of life among people with all rare disorders. It is a multi-part project, and the current survey aims to assess the impact of COVID-19 pandemic on people with a variety of types of rare disorders/diseases, including access to healthcare and information and psychosocial support.

What will we do with study findings?
We will share the overall results with you and communicate them to decision-makers including rare disorder organizations, healthcare professionals, policy makers, and researchers to ensure the needs of people with rare disorders are met during the pandemic.

Who is eligible to participate?
You must be an adult or the age of majority in your state, be able to communicate in English, and have any rare disease or disorder or undiagnosed rare condition. Caregivers who do not have a rare disorder themselves are NOT eligible to participate at this time. A disease is generally considered rare if it affects fewer than 200,000 affected individuals in the United States or fewer than 1 in 2,000 in Europe. A list of rare diseases can be found here: https://rarediseases.info.nih.gov/diseases/browse-by-first-letter. Because rare disorders are discovered and prevalence estimates change frequently, you may participate even if your disorder does not appear on the list.

Who is the principal investigator?
Kathleen Bogart, PhD, Principal Investigator, Associate Professor of Psychology at Oregon State University, studies psychosocial support and healthcare access for people with rare disorders, has a rare disorder herself, and is an advocate. Contact her at kathleen.bogart@oregonstate.edu or 541-737-1357.

PLEASE HELP SPREAD THE WORD!
To ensure that results reflect the diversity of the rare disease community, it is crucial that as many people living with a rare disease as possible take part. Please share this with others who may be interested in participating.

What would I do as a study participant?
If you choose to participate, please follow the link to take the 20-minute online survey: https://oregonstate.qualtrics.com/jfe/form/SV_3lor9lBREFvD4QB
(OAA) provides information and support to parents and professionals dealing with a set of inborn errors of metabolism collectively called organic acidemias. The OAA is a volunteer organization registered with the IRS as a 501(c)(3) non-profit corporation. Donations to the OAA are tax deductible. OAA publishes a newsletter three times a year, hosts a Google Group for information exchange and maintains a website and Facebook page. Services are funded by corporate and individual donations. Annual membership donation of $25 (US) and $35 (international) plus $5 for the family roster is requested, but not required. Our 501(c)(3) non-profit status qualifies OAA for United Way donations through their write-in option. If there is a write-in option, just write “Organic Acidemia Association” in the blank line on your pledge card.

Donations can also be made at OAA’s website through the “PayPal” and the “Network for Good” option.

- The information contained herein does not necessarily represent the opinions of our Board of Medical Advisors or Board of Directors
- Letters and photographs sent to OAA become the property of OAA and may be used or edited at the discretion of the OAA staff.
- Names or information will be kept confidential only if specifically requested in writing
- This newsletter does not provide medical advice. You should notify your health care provider before making treatment changes.