The HCU *Herald*

HCU Awarness Month!

More HCU News Inside

<u>October is HCU Awareness Month!</u>

- HCU Awareness T-Shirt
- HCU Awareness Month Activity List
- HCU Awareness Bingo
- Click Campaign Recap

<u>Events</u>

$20,000 Anonymous Donor Match is Back!

HCU HERO:
ANNA FROM VERMONT

How was your child diagnosed?
It was during winter and spring 1984-1985. After a harrowing four months of denial by the local MD (who'd delivered Anna) that anything was abnormal about her listlessness, failure to respond to feeding attempts, and blue pallor, even his suggestion that Paul, and especially I, were suffering from Munchausen-by-proxy syndrome since I was a "distraught late prime-imp" who asked whether my newborn baby might be having seizures, we had tests run at the larger regional medical center where MDs were equally flummoxed. "Go home and wait," they said. So we asked a medical friend to help with ideas, and she had us order the same tests run on our daughter at the smaller hospital where she had been born. The technologists and nurses there said, "We don't know what's wrong, but there definitely is a serious problem; these parents are upset for valid reasons."

We went back to the bigger hospital for more tests, but this time with respected local medical opinion behind us; our daughter, now almost 5 months old, was then readied for Boston Children's Medical Center. No ambulance was available for the transport to Boston, so we drove her there ourselves. Very soon after our arrival we were picked up by a promising young resident, Dr. Holmes Morton, whose research into inborn errors of metabolism (IEM) led him to compare Anna's situation with babies he'd read about--a boy in Canada and another in Australia who presented in similar ways. He called in a senior associate, a world-renowned physician named Harvey Levy, who'd written seminal pieces about IEM. This doctor was gentle and supportive, not accusatory and judgmental. He sat with us in our initial interview and hand-wrote 14 pages of notes from our observations of Anna's failure to thrive, seizure-like behaviors, and general disinterest in anything around her.

The young resident and the world-class MD conferred with other physicians and, after new and different tests were run and a megaloblastic anemia was found (a lethal development), agreed that Anna was suffering from an IEM, probably in the family of vitamin B12 (cobalamin) disorders. This had been the young resident's conjecture, even before those tests were run. Anna was studied in-hospital, given injections of hydroxocobalamin that made her scream, and given more tests. My sad little pincushion baby! A very short time later, Dr. Levy noticed Anna's slightly improved blood values and said, "I think we can do better for Anna." He switched the hydroxyCbl for injections of methylcobalamin (MeCbl), explaining that it is more bioavailable than the "hydroxy," as it's reduced by one step, a simpler substance, and easier for a baby's body to use.

We asked many questions and Dr. Levy kept up. He realized that teaching us about our daughter's problem was better than having us sit and stew in a waiting room. We were sent home with Anna, but with things to do. When we asked for reading ,Dr. Levy gave us assignments. When it was clear that Anna's metabolic protocol needed to be supplemented with other substances that are hard to find in the US, he let us feel like research partners by having us hunt for these metabolites in a variety of different states and European countries. Some of them, we found and obtained. And Dr. Levy had contacts in Japan, where the best MeCbl then available in the world could be obtained; he and his Japanese contact arranged for a steady supply for Anna. (It later became more widely available, and much more affordable, in this country.)

A few weeks into Anna's treatment, the seizures I had seen after her birth returned, and even more intensely. We went back to Boston Children's Hospital for more tests, resulting in a diagnosis of infantile spasms with hypsarrhythmia, a severe and damaging seizure disorder. Through the summer 1985, we tried a conventional anti-seizure medication, supervised by phone discussions by Dr. Holmes Morton. By September, when that strategy produced no results, Paul brought Anna back to Boston Children's for the start of ACTH treatment and coaching for home administration. During that time, Paul met another physician, a pediatric neurologist with a developmental background who later became our local metabolic advisor--James Filiano, MD. The ACTH wasn't a total cure but did stop the deathly hypsarrhythmia. After that, Anna began to grow. She gained weight and strength, color in her cheeks, and at least passing interest in things and people around her.

When Anna was eighteen months, we asked Dr. Levy to arrange an MRI for her; the results were disturbing. Her brain lesions were many, and her optic nerve had only a trace of the myelin sheath normal for that age. But Dr. Levy assured us that if we held to Anna's medical regimen with care and precision, we would see improvements. The second MRI at 36 months was dramatically improved. Anna was clearly able to see to some degree, and her coordination had been improved by the treatments. The MRIs were taken and became part of the diagnostic data used by a former student and then colleague of Harvey Levy's in Montreal, the scholarly David Rosenblatt, himself a Cobalamin researcher and diagnostician. At this time, Dr. Rosenblatt completed his differential diagnostic study of Anna's cultured fibroblasts, in his labs at McGill, determining her problem to be Cobalamin G disorder.

How has the diagnosis affected you, your family and relationships?
It totally changed the dynamic of our lives. Instead of becoming a nice, upwardly mobile family with a lively social calendar full of playdates, PTA fundraisers, and vacation parties, we circled our wagons around our daughter, concentrating on her development through steady refinements of her medical protocol and her work with the best therapists available for all areas in which she was affected: physical, occupational, sensory, visual, speech, and musical. Some strategies worked; many did not.

Then came her education, starting with day care and nursery school, followed by Waldorf and recreational camp, then special education locally. We did not want to have her schooled in a distant location where we couldn't monitor her learning and support the process and the practitioners. Her public elementary education was a huge challenge to the local teachers, who did the best they could, Anna's disabilities being significant--no speech, uncoordinated gait, minimal fine motor control, visual problems, and more. Her secondary-level learning at the independent Academy in our town was more successful. She had dedicated teachers with some background in developmental disabilities and autism, along with more inclusive school practices.

We provided Anna with developmentally appropriate summer programs for years until school funding started for such things. Fortunately, our state's agencies have been increasingly supportive. But for a long time--and now in the pandemic, it's pretty much mostly us. Anna is back home with the old folks. And now in the Age of COVID-19, who really has the sort of social life they want? Along this journey, our lives have been enriched by the great number of phenomenal people who have helped us and Anna.

What are some of your successes with the disorder?
Success won't be attained until we've cured her and helped her overcome the neurologic challenges she deals with daily. We continue to work with Anna through medical nutrition and physical and communication development. It's slow going, but we have a wonderful team.

What are some of the challenges you have faced?
Finding and affording meds and medical nutrients; learning to read and understand medical and developmental literature; finding effective therapists for our daughter; finding programs to stimulate, teach, and involve our daughter in learning how to move, behave, interact with others and with animals, and manipulate materials; teaching her and helping others to teach her skills at home and in the community, working with teams of doctors and educators to maximize Anna's development. She has needed special strategies to help her overcome muscle contractures that started in her earliest days.

Well into Anna's adulthood, at the recommendation years before by Dr. Harvey Levy, we ordered an MRI for Anna to determine the reason for her unsteady gait; a congenital tethered cord was discovered. Though the "fix" for this problem is of questionable effectiveness, the tethered cord explains her deeper problems and we are accommodating that reality. Along the way and later, including now, maintaining our extended family connections and friendships with a minimum of travel and expense. Taking vacations that typical middle-income Americans would expect to enjoy has been extremely difficult, so these days we don't even try. We live in a vacation area, which is good enough for us.

What words of advice would you give to newly diagnosed families?
Decide at the outset whether or not your love for your partner and your child can be respected before anything else. If it is a powerful enough factor in your life, it can get you through almost anything. In the darkest, pre-diagnosis days of our parenthood, Paul and I shared a motto: "NO MATTER WHAT..." The rest of that statement could be "I love you" or 'we love our baby" or "we will work for as long as we last to help our family grow," etc. It became a quick way to affirm our dedication to helping Anna, and of our mutual love.

Despite all of these challenges, Anna has grown up as a person who has unexpected strengths. She is learning to type on electronic keyboards and has shown herself to be cognizant of the world around her, observant of people's needs and motivations, and capable of wit, metaphor, irony, and sarcasm. She can be funny as the dickens. And she is kind-hearted. We are proud of our daughter. That's her success, though I suppose we can say we share it.

HCU NETWORK AMERICA
IS LOOKING FOR STATE AMBASSADORS

Looking for active and outgoing members of the HCU community

What does an ambassador do?

Ambassadors...

- *Connect with local HCU families*
- *Share their story*
- *Advocate and raise awareness for HCU*
- *Amplify and support our mission*
- *Help fund-raise*

Get involved today! Contact Danae'
dbartke@hcunetworkamerica.org

BECOME A STATE AMBASSADOR FOR HCU NETWORK AMERICA

HCU Network America

Will you be our next HCU patient Hero?

Tell us:

- How you or your child was diagnosed?
- How has HCU affected you, your family and relationships?
- What are some of your successes with HCU
- What are some of your challenges you have faced?
  - How have you overcome them?
- What words of advice would you give to newly diagnosed families?
- For other patient stories, visit: https://hcunetworkamerica.org/patient-stories
- Email your story to: info@hcunetworkamerica.org

October 2020: HCU and You: *Ask Methia*

Dear Methia,

...then Comes Planning, then Comes the Babies and the Baby Carriage

My partner and I have been talking about starting a family. We are trying to be responsible and have been talking about all of the things we should do to prepare for trying to conceive. Of course, this includes making sure that I am set up for a healthy pregnancy with homocystinuria. I don't know anyone with homocystinuria who has become pregnant or had a child yet, and I feel like this isn't something my parents talked with me about when I was a teenager. Do you have any suggestions?

Sincerely,
HCU Mama (In Waiting)

Dear *HCU Mama,*

Congratulations! Deciding to try to grow your family is a very exciting time, and also a little stressful for most people. Making sure you are in optimal metabolic control for both you and your baby adds an extra stressor on top of that. Pregnancy was once thought to be contraindicated for many inborn errors of metabolism. As therapies for patients have improved over the years and more patients are thriving as they reach reproductive age, this has preconceived notion has been proven untrue. Here are some things to keep in mind as you start your preconception process:

1. **Inform your clinic about your plans as soon as possible.** Your metabolic physician will want to go over recommendations and expectations, and may want to optimize doses of your vitamin cofactors or medications. This might be necessary if your weight has changed since your last visit, or your prior homocysteine levels or methionine levels were outside of goal range.

2. **Your metabolic clinic will ask that you follow with them very closely – possibly even weekly.** Your caloric and protein requirements will increase during pregnancy. In order to assure proper fetal growth and keep you in good metabolic control, you will need frequent weight checks and lab work. Your team will assess your nutritional needs as they rapidly evolve, and prescribe you the right amount of calories and protein (from both food and formula) to keep both you and your baby healthy. If you are not already tracking your caloric and protein intake daily, now is definitely the time to start!

3. **Your clinic will help you with a plan for a healthy delivery.** Childbirth places a significant amount of stress on the body, and having homocystinuria places you at higher risk for delivery complications. Your team will make a protocol for you that will include dextrose-containing IV fluids, as well as specific medications to prevent blood clots (before, during, and after delivery).

While still considered a high-risk pregnancy, there are patients with homocystinuria who have had healthy babies and achieved good metabolic control thanks to preventative management and very close follow-up. Having a healthy baby makes all the hard work to have good metabolic control worth it!

Sincerely,
Methia

START TO FINISH DELICIOUS!

Click picture or name for recipe

APPETIZER

ZUCCHINI FRITTERS

MAKES 6 SERVINGS
SERVING SIZE: 1 FRITTER
PROTEIN PER SERVING: 0.6 G
CALORIES PER SERVING 71

SOUP

SLOW COOKER VEGGIE CHILI

SERVING SIZE: 8 OZ.
PROTEIN: 2.3 PER SERVING

ENTREE

PUMPKIN PESTO PASTA

MAKES 1 SERVING
PROTEIN: 2.5 G
CALORIES: 553

DESSERT

SOFT PUMPKIN COOKIES

MAKES 12 SERVINGS
SERVING SIZE: 1 COOKIE
PROTEIN PER SERVING: 0.2 G

HCU Network America HALLOWEEN CANDY GUIDE

FREE!

- Airheads
- Bottlecaps
- Candy Necklaces
- Dots
- Fruit Runts
- Fun Dip
- Gobstoppers
- Hard Candy
- Jolly Ranchers Hard Candy
- Laffy Taffy
- Lifesavers
- Mike & Ike
- Nerds
- Pez Candy in Dispenser
- Pixy Stix
- Ring Pop
- Salt Water Taffy
- Smarties (U.S. version)
- Sour Patch & Sour Punch Candy
- Suckers/Lolly-Pops/Dum Dums
- Swedish Fish
- SweeTARTS classic

LOW PROTEIN

LESS THAN 0.5g PROTEIN

PER PIECE

- Licorice, Bites
- Marshmallow Ghosts
- Sugar Daddy, Junior
- Tootsie Roll Midgees
- Tootsie Roll Pops, Caramel, Chocolate, or fruit flavors
- Hershey Hugs
- Hershey Kisses
- Kraft Caramels
- Red Vines Black Licorice Twists
- Red Vines Original Red Twists
- Rolo Caramels in Milk Chocolate
- Twizzlers Licorice Twists
- Twizzlers Strawberry Twists

PER PACKAGE

- Candy Corn
- Jelly Beans, all flavors
- Skittles, all flavors
- Sprees Chewy Candy
- Starburst Fruit Chews, all flavors
- SweeTARTS Chewy
- Sugar Babies, Fun Pack

HIGHER PROTEIN 0.5g+

MINI CANDY BARS

$100 Grand, 3 Musketeers, Almond Joy, Baby Ruth, Butterfinger, Hershey's Minis, Kit Kat, Milky Way, Mounds, Snickers, Twix

PER PIECE

- York Peppermint Patty
- Reese's Peanut Butter Cup, miniature

PER PACKAGE

- Haribo Gummi Bears
- Jolly Rancher Gummies
- Junior Mints, regular size
- Life Saver Gummi Savers
- M & M's, fun size
- Malted Milk Balls
- Milk Duds, 13 pieces
- Raisinets, 1.58 oz. bag
- Sugar Babies, regular size
- Sugar Daddy, regular size
- Whoppers

IDEAS FOR HANDLING HALLOWEEN CANDY

- Set aside higher-pro candy for the "switch witch" who comes & brings a present on Halloween night
- Trade in higher-pro candy at the dentist or donate it at local firehouses or other organizations
- Trade higher-pro candy with friends and siblings for lower-pro options

CREATED FOR HCU NETWORK AMERICA BY

NATIONAL PKU News

All data is based on values from HowMuchPhe.org. HowMuchPhe.org is a service of National PKU News. Free trials of HowMuchPhe.org are available. Visit the site for details.

Taste Connections, LLC

(310) 413-6499 | tasteconnect@verizon.net
Tasteconnections.com

Family Owned and Operated Since 2003

- Made to Order
- No artificial preservatives, colors or flavors

Mixes: The more you buy, the more you save (MUST ORDER BY OCTOBER 10TH TO RECEIVE DISCOUNT)

- Buy 2 bags, take $1 off each bag, buy 3 bags and take $2 off each bag
  - TC- Bread Mix - $24.50 per bag (base cost)
  - TC-Multibaking Mix - $25.50 per bag (base cost)
  - TC-Brownie Mix - $16 per bag (base cost)

Loaves of Bread:

- Seasonal Quick Breads for Fall -$12.30 per loaf
  - Apple Craisin Bread
  - Pumpkin Spice Bread
- Cinnamon Swirl Bread - $11.50 per loaf
- Freshly Baked Sliced White Bread -$11.50 per loaf

Rolls/Buns: $13.75 per package

- Focaccia/ Pizza Bread - 4 individual pieces per package
  - Great for a pizza base with your favorite low protein cheese and veggies!
- Herb Sub Rolls - 4 individual pieces per package
- Hamburger Buns - 8 individual pieces per package

Goodies:

- Cookies - $11.50 per bag
  - Seasonal: Apple chunk/cinnamon cookies
  - Year Round: Chocolate Chip, Double Chocolate Chip, and Strawberry Jam
- Caramel Tapioca Party Mix - $9 per bag
  - Caramel Tapioca, Craisins, dried sweetened pineapple and apple, candied lemon, white "chocolate" bark

GO BLUE FOR HCU

October is HCU Awareness Month

This for HCU Awareness Month is Go the Extra Mile for HCU. How do you exactly go the extra mile for HCU? You raise awareness, you fundraise, you advocate and help amplify the messages of the HCU community.

October is packed with exciting things!

2020 HCU Awareness T-Shirt:
Get your official 2020 HCU Awareness T-Shirts. While you wear your shirt, you help raise awareness for HCU, and by purchasing you are helping raise funds to benefit HCU Network America's Educational Program Fund. So order your shirt now so you have it in time for HCU Awareness Month!

Click the link to purchase - https://bonfire.com/go-the-extra-mile-for-hcu-homocystinuria

HCU Awareness Bingo:
Want a chance to earn some HCU Awareness Swag?

- We will be giving away a book, wrist bands, awareness socks, and other fun prizes!
- Bingo pieces will be shared each day during the first full week of HCU Awareness Month on our social Media channels – you must tag us and use the hashtag #HCUAwareness2020
- Connect 5 in a row – horizontal, vertical, or diagonal to win! Each Bingo is a new chance to win.

OCTOBER IS HCU AWARENESS MONTH

HCU Awareness Calendar
Want to do more to raise awareness for HCU? Check out our fun online activities you can take part in to help those understand more about HCU. **Additionally, look out for our fact of the day**

Share a fact from our fact calendar, or complete an activity each day and use #HCUAwareness2020 and earn a chance to win a free HCU Awareness t-shirt!

- ☐ Change your social media picture to the HCU Awareness Ribbon
- ☐ Start a HCU fundraiser
- ☐ Share an infographic about HCU
- ☐ Share a patient story
- ☐ Share your diagnosis story
- ☐ Challenge your friends to the same amount of protein and three normal protein shakes a day #ToastTocHCU
- ☐ Share a pic of an item that has the same amount of protein you can have
- ☐ Share your daily diet record –completed
- ☐ Share a low-protein meme
- ☐ Share your favorite low protein recipe! Bonus if you cook it and share a pic
- ☐ Dining out, low protein style. Where do you like to eat?
- ☐ Share a pic of what your grocery store haul looks like
- ☐ Real cost of HCU: Grocery Cost Comparison #Medical Nutrition Equity Act, or share some patients with HCU require injectable B12. B12 on average is $300-400 a month and most insurance companies don't cover it!
- ☐ Share a picture or video capturing all the medication you take (this includes formula for those who need it).
- ☐ Share a picture of your first pair of glasses, or a device that helps you navigate or communicate due to lack of vision
- ☐ Share something you wish people understood about HCU
- ☐ #HaikuforHCU—Write and share a Haiku describing life with HCU
- ☐ Wear jeans for your rare genes #ItsInOurGenes
- ☐ Wear your HCU Shirt and share a pic online—#HopeConnectsUs
- ☐ #GoBlueforHCU
- ☐ #HCUAwareness post in a public place
- ☐ Share with a stranger what HCU is and why it's important to you
- ☐ #Create4Cure—Create a work of art that brings awareness for HCU— can be a song, dance, a painting—get creative!
- ☐ #High5forHCU—List 5 ways HCU makes you a stronger, better person
- ☐ All states test for classical HCU, but many are still missed
- ☐ Share a picture of you and a HCU buddy! Or tag a friend who is of great support
- ☐ #FacesofHCU—Share a picture of you saying, I am one of the 1 in 200,000 people with HCU
- ☐ #Hope4HCU—Share 4 things that give you hope and encouragement
- ☐ Share the HCU timeline—if you know other facts, let us know!
- ☐ Cutting Edge of HCU: Share about a therapy that is in the works!

To find additional information and resources, visit:
https://hcunetworkamerica.org/hcu-awareness-month/

How Can Zebrafish Help Researchers Understand Metabolic Disorders In Humans?

JENNIFER SLOAN PHD, MS AND CHARLES P. VENDITTI MD, PHD

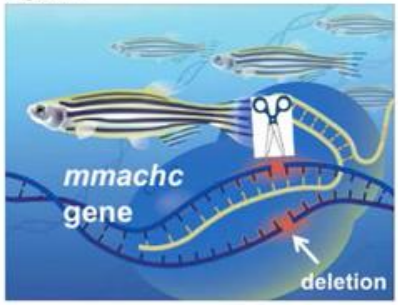
Since our last article update, we continue to study hereditary methylmalonic acidemia and cobalamin disorders in both in the laboratory and the clinic. As many of you know, we have been actively studying one of the cobalamin disorders called combined methylmalonic acidemia and homocystinuria cblC type 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.

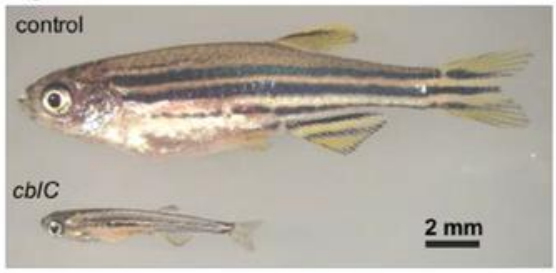
Figure 1



Modified from: https://www.genome.gov

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.

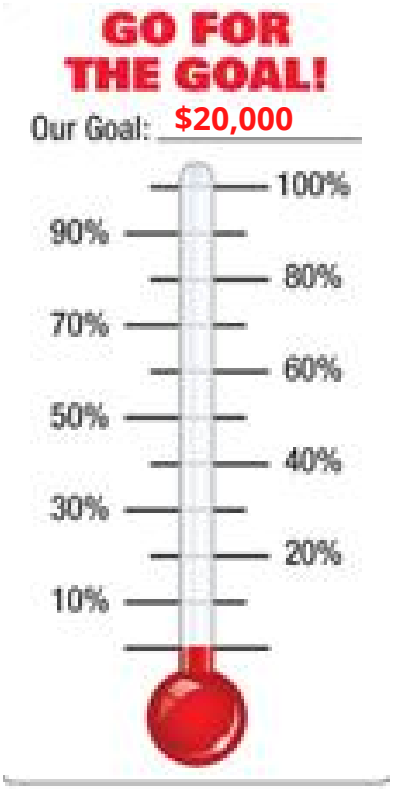
Figure 2



https://doi.org/10.1093/hmg/ddaa044

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.

$20,000 DONOR MATCH IS BACK!



That's right, you heard us right! Thanks to an anonymous donor, any **funds** you help raise from October through December 31, 2019 **will be matched up to $20,000!**

We are asking every patient and family to help us raise funds for homocystinuria. Set up a Facebook Fundraiser or host your own alternative fundraising event and invite your family and friends to participate. Alternatively, they can donate directly to HCU Network America.

Have an idea for a fundraiser, but not sure how to get started? Let our fundraising committee help you get started

Email info@hcunetworkamerica.org and we will connect you!

15,000 Companies Match Gifts....

Does Yours?

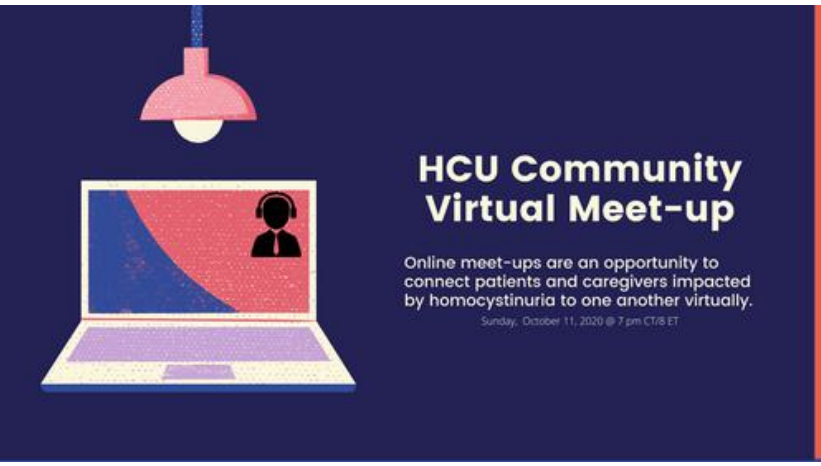
Here are the steps to see if your company matches gifts!

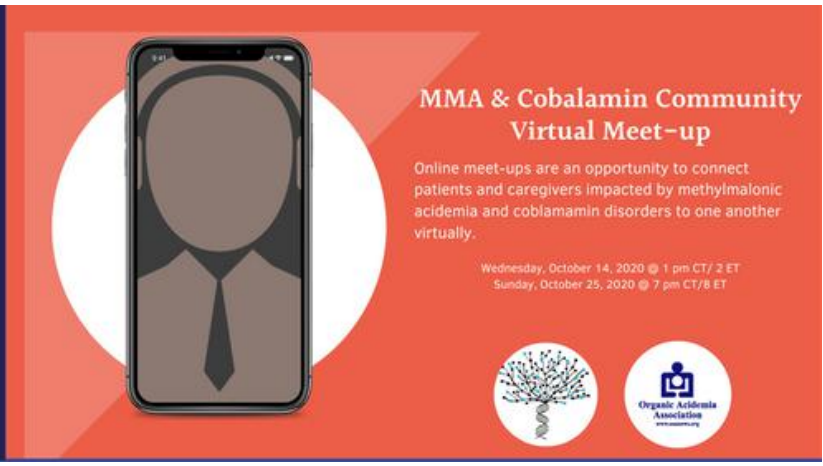
EVENTS

Come check out our Virtual Homocystinuria Meet-ups!

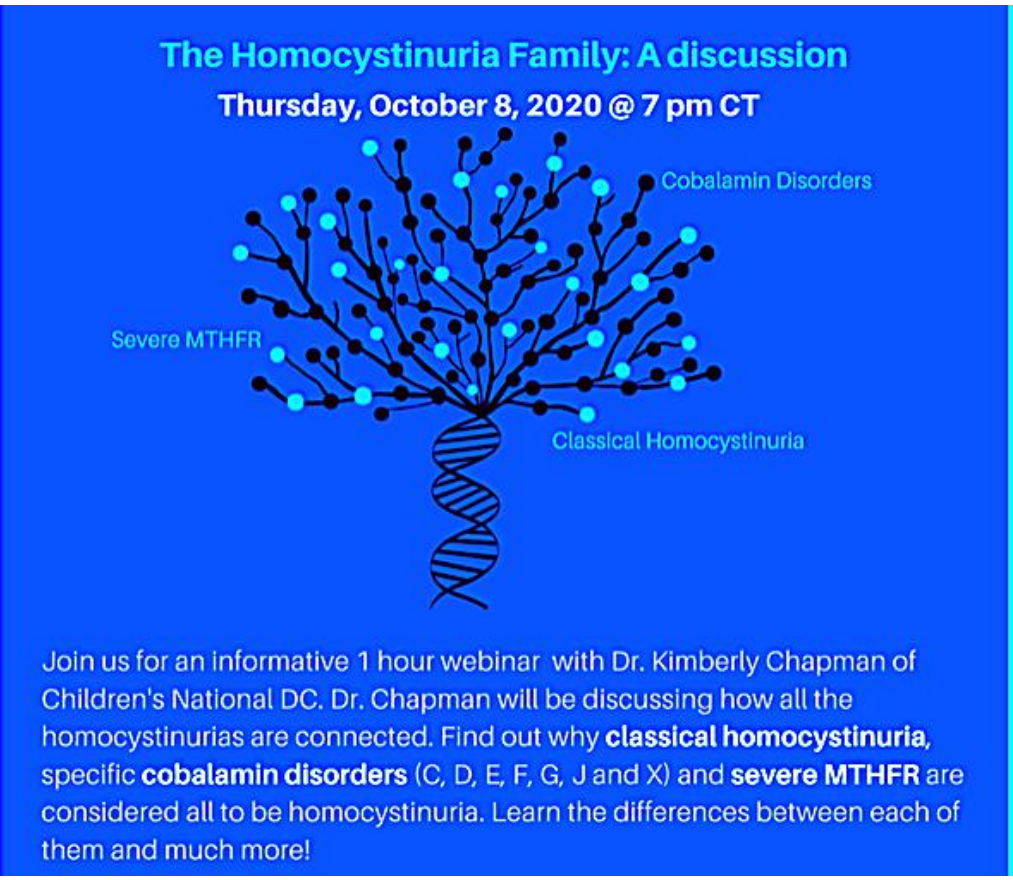
Join our virtual meet-up for a chance to meet, connect, and learn from other patients and caregivers who are facing similar challenges. Whether it's navigating adherence issues, insurance, clinic visit, or life transitions, you are not alone.

Register now at: https://www.eventbrite.com/o/hcu-network-america-30163980100





Come take a deeper dive into understanding Homocystinuria - Attend our webinar!



Dr. Kimberly Chapman is an attending physician in Genetics and Metabolism at Children's National in Washington DC, United States. She specializes in taking care of all ages of individuals with defects of methylation, homocysteine and propionate metabolism. She currently divides her time between clinical responsibilities, clinical research including several patient registries and other clinical trials, and a basic science laboratory

Register now:
https://bit.ly/HCUWebinar





Rare Bears for Kids with HCU

RARE science

Accelerating Cures for RARE Kids

We are thrilled to announce that the Rare Bears for HCU Campaign in partnership with Rare Science has been re-opened!

To enroll in the RARE Bear Program and to request a RARE Bear, please click the link and complete the form: https://www.rarescience.org/hcu/

- Those who have already received a bear, are not eligible
- Date for gifting will be announced later
- You will **not** receive a confirmation email or be notified when your bear has been shipped

Click Campaign Recap

During the last full week of September, Recordati Rare Diseases created a click campaign that benefited for the HCU community. For every "click" Recordati Rare Diseases donated up to $5 to HCU Network America. Thank you Recordati for partnering with us to raise awareness and funds for the homocystinuria community! **This year we had 639 clicks** – compared to the 525 clicks from last year!

GO THE EXTRA MILE FOR HCU, RACE RECAP

Thank you to the 95 racers and countless people who donated to make our second virtual race a smashing success! The first week started off very strong. We had 870 miles logged and 3 racers cross the finish line! First place in our cycling division went to Allison T. of Team Aeglea, and in our running division first place went to Laura P., and second place went James S. of Recordati Rare Diseases and In addition to the strong lead by racers, Brooke's Blazers, Recordati Rare Diseases and The Bartke Ruff Ruffs, Ellie's Entourage and South Florida Strides had already passed our fundraising goal and had collected $5,815 in donations!

As week two started, the momentum picked up and racers logged another astounding 1,016 miles! As the competition began to increase, we started to see racers cross the finish line and 12 more racers crossed the finish line. As week two came to a close Ellie's Entourage added another $550 to our fundraising goal, for a total of $6,365

The second leg of the race many new racers entered the leaderboard and 2,030 miles were added to the total completed. With that extra competition another 9 racers crossed the finish line. With only 21 of 95 registered racers having crossed the finish line, those watching the race became slightly skeptical if everyone was going to finish with so many mile left completing.

With only a few days left to go, our racers picked up their pace. In the end, 65 racers logged over 4,888 miles and 44 crossed the finish line. With time ticking donations continued to come in making for a final total of approximately $9,492! This truly was an amazing event. Thank you to all our amazing racers and donors for making this an
exceptional event!

Meet our Fabulous Fundraising Teams!

Brooke's Blazers - $3,150
Our daughter Brooke was diagnosed with Homocystinuria with MTHFR deficiency on 8/14/2020, at three weeks old. Her condition was originally caught by the California Newborn Screening Test. We spent a total of 27 days at Valley Children's Hospital in Madera, CA, while our geneticist worked towards the diagnosis and began treatment. The last 12 days we were solely waiting for the medication to arrive so we could go home, a frustration I imagine many of you know too well. When we heard about the HCU Network and the virtual marathon, we knew that it would be a great opportunity to spread awareness among our family and friends and to raise money for a cause that is now very near and dear to our hearts! We look forward to getting more involved in the community and continuing to raise money and awareness for HCU! - Mike, Erika, & Brooke Hudelson

Ellie's Entourage - $1,515

Entourage walked on behalf of Ellie, 2 year old with classical HCU. We walked not only to show support for Ellie, our daughter/granddaughter, but to raise awareness for the disease. We hope that the funds we raised go to advocating for better treatment options and support for people with HCU. The HCU network has been important to us during our journey navigating how to be the best support for Ellie.

Give Will A Shot - $1,100

Chris and Tara Hummel tag teamed to raise money for the Give Will a Shot team. Chris has run a few marathons in the past, so he was happy to have a month to run 26.2 miles instead of only 4 hours. Tara is a new runner, but did a great job getting her miles in! Both Chris and Tara gain motivation and inspiration from their son Will, who has HCU. He is such a trooper with regards to his diet and treatment. It would be easy for him to say he's had enough, stray from his diet, and give up. However, he never does. Chris and Tara try to duplicate that mentality while running...just keep going even when you want to stop. We've learned that where there's a Will there's a way! Thanks to all of our friends and family who supported us this month on our runs!

Team Recordati Rare Diseases - $1,026

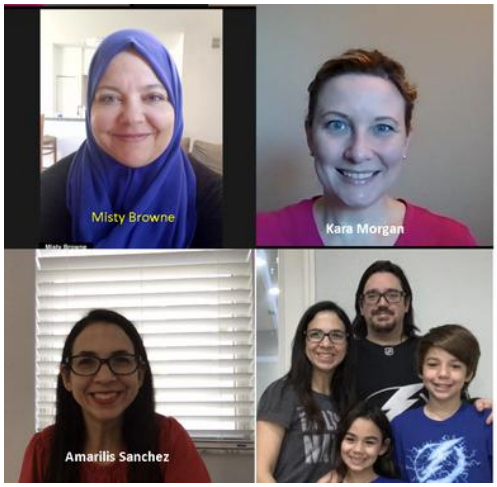
Team Recordati Rare Diseases is a team composed of 35 of their employees. Recordati Rare Diseases is a biopharmaceutical company committed to providing often overlooked orphan therapies to the underserved rare disease communities of the United States. Our experienced team works side-by-side with rare disease communities to increase awareness, improve diagnosis, and expand availability of treatments. Recordati Rare Diseases is proud to support HCU Network America and the Homocystinuria community. For more information, please visit www.recordatirarediseases.com

The Bartke Ruff Ruffs - $1,001

Danae' and Garrett were diagnosed with classical Homocystinuria in 1995 at the ages of 10 and 5 after Garrett's lenses became dislocated. For years they struggled with the low protein diet and dealing with issues related to non-compliance. In 2009, after Danae' suffered a blood clot they were connected with the PKU Organization of Illinois and Allied Disorders, where they were invited to low protein cooking classes and educational events. Danae' loved the support and got involved in their organization. Her time volunteering lead her to realize the gap in support for the large Homocystinuria, which then lead to helping co-found HCU Network America. Danae' feels fundraising is a way to give back to the community that has provided so much for her!

Grayson's Gleeful Galloping Gang of Goons - $800

I have been an avid runner my whole life and enjoy the competitive side of racing. While I am not very fast anymore, and often run with a child or two in tow, I still enjoy setting goals and working hard to accomplish them. While I am usually the only one who participates in races, our whole family is very active and loves to be outside. Signing our family up to participate in the HCU race was driven by my desire to not only raise awareness for Grayson's disorder and fundraise; I was hoping it would help our family set a goal and accomplish it together. Since Grayson has been born, our family motto has been "divide and conquer." Due to COVID and the safety precautions we have taken to keep our family safe, we rarely go out and do anything together as a family anymore. Not to mention the time and energy it takes to ensure Grayson takes all his medication (some days are easier than others). However, participating in the race together, walking and running together and supporting each other as we accomplished our goal has been a bonding experience for our family. I have also been incredibly blown away by the support and love from our extended family and friends. While my initial goal to fundraise was $100, we have currently raised $780! I'm grateful for the opportunity we have had this month to focus our family and spend time together.



South Florida Strides - $350

South Florida Strides is a team created by the University of South Florida metabolic team. We have patients with homocystinuria and we wanted to participate to raise awareness. Misty Browne is our metabolic nurse, Kara Morgan is our nurse practitioner, and I am the metabolic geneticist.

Jaime FT is my husband, Jaime FS and Anasofia FS are our kids and they wanted to participate too!

Team KJ - $300

Team KJ is composed of Kristin Rapp Clubbs and her husband JP Clubbs. We accepted the challenge of the Go the Extra Mile for HCU Virtual Race both to bring awareness to the rare disorder Kristin was born with - Homocystinuria (HCU) - and to keep active during the month of September. Making the commitment to walking a minimum of 26.2 miles over the course of a month was a great way for us to spend some quality time being active outdoors (safely socially distancing of course!). We hope to keep the momentum going as we head into HCU Awareness Month - which starts on October 1st!

HC&U HOSTED BY BEN AND LINDSEY MASSENGALE

– PATIENT STORIES
– RECIPES
– RESEARCH
– TIPS FOR HCU MANAGEMENT

FIND US APPLE PODCASTS AND STITCHER!
WWW.INSTAGRAM.COM/LOWPROBITES
WWW.FACEBOOK.COM/HCUPODCAST
WWW.TWITTER.COM/HCUPODCAST

Focused on the Few™...

At Recordati, we focus on the few - those affected by rare diseases. They are our top priority and at the core of everything we do. Our mission is to reduce the impact of extremely rare and devastating diseases by providing urgently needed therapies. We work side-by-side with rare disease communities to increase awareness, improve diagnosis and expand availability of treatments for people with rare diseases.

Recordati Rare Diseases is proud to support HCU Network America in their commitment to people living with HCU.

RECORDATI RARE DISEASES GROUP
Focused on the Few™

www.recordatirarediseases.com/us
@RecordatiRare
NP-RRD-US-0162

HEARTS ENTERAL, LLC

Re: Introduction to Hearts Enteral – DME – DBD program.

We would like to introduce our company, Hearts Enteral, LLC, which is a new DME supplier specifically designed for enteral nutrition and medical foods.

Hearts Enteral is a program/DME provider to help patients afford and distribute their formula. We are a unique supplier that is different from the typical suppliers. We work our magic!

We are a team of experienced certified medical billers (CBCS) that work specifically for medical foods and enteral nutrition reimbursement. Our goal is to look for new ways to help the rare genetic disease community afford and stay on diet.

We understand the complexity of other suppliers that drop or don't accept a certain product order based on cost vs. reimbursement (low reimbursement). This could hurt manufacturer's profits and patients health.

It's a sad loss! Don't you agree?

Based on the ongoing complexity with medical food distribution, we have developed a program that manufacturers and patients are sure to appreciate and value – **DBD Your Way (Direct Billing and distribution).** This program will help alleviate the manufacturer's time and maximize their revenues for medical foods/enteral nutrition products. In addition, DBD Your Way will help patients receive the appropriate coverage and distribution in a one stop shopping area.

No more searching for suppliers! Or at least as a last resort just to keep it moving.

We are asking manufactures to join Hearts Enteral' s new program "DBD Your way" to help patients receive their products timely, as well as get the proper coverage and reimbursement.

All we have is each other to help one another! This unique program offers:

- Direct billing services and appeals. We won't stop until we get the fair payments.
- Over 10 years medical billing and coding experience specifically for Medical Foods, enteral nutrition and specialty vitamins.
- Product(s) distributions (through the manufacture's warehouse(s) only)
- Hearts Enteral's DBD Your Way program will help maintain patient's dietary medical foods on a regular basis and provide valuable growth benefits to manufacturing companies. **Best of all, less down time!**

How it works, Hearts Enteral contracts with medical food manufacturing companies to help support our program and supply their products.

Consists of: The DBD Your Way program consists of placing medical food orders to Hearts Enteral, LLC either directly from clinics, patients or from the manufactures insurance support programs and we will direct bill to the patient's insurance carriers. The reimbursement would come directly from the insurance carriers and paid directly to the manufacturer. Very simple!

We are already building and started working with manufacturers.

For more information, please contact Raenette Franco or Michael Magzanian at franco@heartsenteral.com or call 973-832-4736.

We look forward to a potential fruitful working relationship in the future!

Sincerely,
Raenette Franco, CEO
CC: Michael Magzanian, CFO

~We donate to non-profit organizations to help change the lives of the rare disease community ~