The HCU Herald

Featuring...

HCU Hero

Mila from Vancouver

November 2022

All things Homocystinuria: patient stories, resources, research, events and more!

HCU HERO: MILA FROM VANCOUVER

Mila's Story

Our daughter Mila was born in October of 2018. She is our little ball of fire, in the most beautiful way! She is feisty, so smart, and she thinks she is so funny. One week after our little fireball was born, her father and I received a phone call that would leave us paralyzed. It was about Mila's Newborn Screening results. Based on her results, it was determined that Mila had "Classical Homocystinuria". It was tremendously difficult for us to process the information. We had just met our beautiful baby girl, and already our minds were racing with every emotion and scary thought. We had no idea what Homocystinuria was and thought we were going to lose our daughter.

After that phone call, we spent over a week at British Columbia Children's Hospital with Mila where an amazing team of doctors, nurses, and dieticians treated her to bring down Mila's levels, as they were quite high. The doctors also helped to come up with a plan to keep her levels down moving forward. The team was not only helping us but also learning more about Mila's condition due to its rarity. Because of how high Mila's levels were, an MRI was suggested to make sure she had no edema (swelling in the brain). This was one of the scariest times for us, as we were terrified of what they were going to find. Once her results came back, we were relieved to find out that she was okay.

Our metabolic team spent day after day with a trial and error approach with her formula and medications to make sure their treatments were going to keep Mila's levels stable. At the same time, they were also consoling us along the way, as they knew how scared we were. She was just so perfect and so small; she truly was a little trooper. As of now, Mila's treatment plan consists of B6, Folic Acid, and Betaine, along with her formula and now monthly blood work. We knew after we left the hospital, that not only have our lives changed but also Mila's. The metabolic team left us with all the tools to successfully manage her condition at home, along with access to them when needed. I was still so nervous about leaving the hospital with this beautiful little baby and I was scared of how I was going to continue her treatment plan without all the support around me.

> ***As parents, the biggest challenge is trying not to worry.***

As parents, the biggest challenge is trying not to worry. Millions of questions ran through our heads: What would Mila's life look like? What about her diet? Will she have development problems? Is this condition going to cause other health problems? How will she feel when she is at a birthday party and all the other kids can eat the birthday cake and all the treats and she cannot? And one day, if she chooses to have children, what is pregnancy going to be like for her? It was just never-ending questions and a feeling of uncertainty for her father and me.

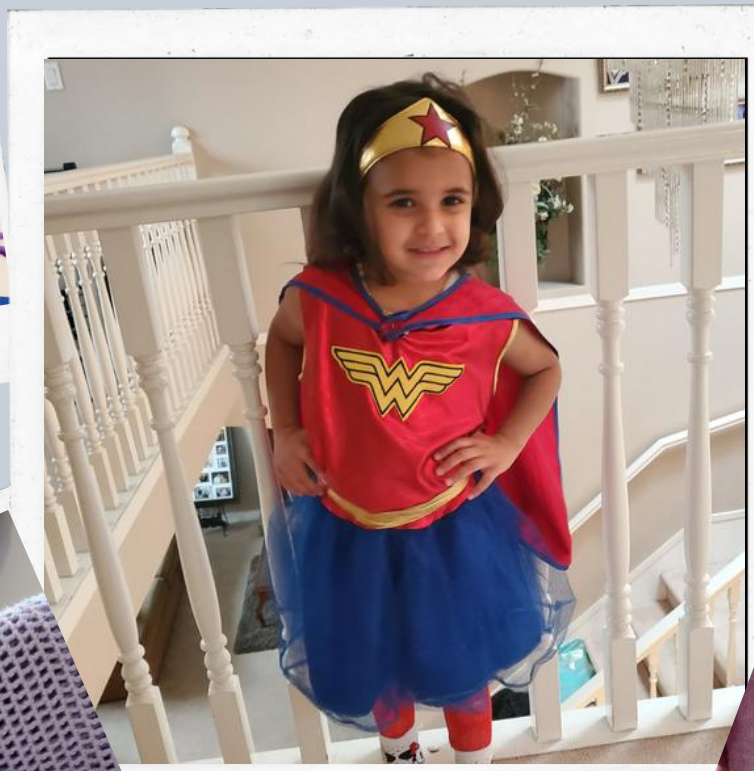
Another big challenge that we began to face was that Mila was becoming quite the picky eater. First, with all her restrictions and now on top of everything, she was being so picky. We were constantly so afraid that she wasn't getting enough nutrients and I became obsessed with food and feeding her all the time. However, despite all of our worries, we were always reassured by her doctors that she was doing well.

Once I had a chance to stop and reflect, I realized that moving forward, I would need to take each day one day at a time. I realized that I needed to be grateful that her condition was caught early on and that we live in a time where there are many different options for food and other various resources, including all of the amazing support that we have had. We even had a development coach come and see her every 3 months to make sure she hit every milestone, and she did!

For parents who have children with HCU, I feel that the best way to navigate through this is to reach out to people who are going through the same or similar situations. Having a support system helps so much. I know it helped us with feeling anxious at times, knowing we could talk to someone who knew what we were going through. This made us feel a lot better and made us feel like we weren't alone. You can learn a lot from each other, whether that means just having a shoulder to lean on or sharing resources with one another.

> I realized that I needed to be grateful that her condition was caught early on and that we live in a time where there are many different options for food...

Our goal for Mila is to teach her independence and my goal as her mother is to teach her to be a strong woman who will be ready to take care of herself one day and not let this condition stop her from achieving her goals and aspirations in life. I don't want Homocystinuria to define her. She is such a strong-willed little girl and we never want that fire in her to die, and as her parents, we will never allow that to happen. We watch as her teachers adore and praise her, we watch her play her little heart out on the soccer field, we watch her pretend to be a mermaid during her swimming lessons, and it is within these little moments that we see who she is, and we see who she will become.



This HCU Hero story was written with love by Luvy, mom of Mila.

Pâté

***Yields 9.8 servings* | Serving Size: 1 oz. (28g) | Protein per serving: 0.5 g**

Ingredients:

- ▯ 1/4 c Diced Raw Onions (10 *mg*)
- ▯ 2 clove(s) Garlic, minced (10 *mg*)
- ▯ 88 g White Button Mushrooms, about 8 ounces, chopped (75 *mg*)
- ▯ 2 TBSP Olive Oil
- ▯ 2 TBSP Bragg's Coconut Aminos (2 *mg*)
- ▯ 4 fl.oz. Red Wine, optional (4 *mg*)
- ▯ 1/2 tsp Bouillon, Beef 1/2 cube, crushed
- ▯ 1 TBSP Chopped Fresh Rosemary (3 *mg*)
- ▯ 1 TBSP Butter (6 *mg*)
- ▯ 1/4 c Daiya Cream Cheeze, Plain, softened (56 *mg*)
- ▯ 1/2 tsp Balsamic Vinegar
- ▯ 1/4 tsp Salt
- ▯ 1/2 tsp Black Pepper (4 *mg*)

Directions:

1. Preheat olive oil in a medium skillet over medium heat. Add onions and sauté until aromatic, about 3 minutes. Add the mushrooms and garlic. Sauté until the mushrooms are browned, about 5 minutes.
2. Add the coconut aminos to the cooked mushrooms. Cook until the aminos is absorbed. Add the red wine, crushed bouillon, and chopped rosemary. Cook until most the red wine is absorbed. Remove from heat and add the butter. Stir until melted. Set aside to cool for 5 to 10 minutes.
3. Add the mushrooms to a food processor. Pulse, until desired texture is reached. You can leave it a little chunky, or smooth like a spread. Add the softened cream cheese and balsamic vinegar. Pulse a few times to combine. Add salt and pepper as desired. Serve immediately or refrigerate and serve chilled.

Note: You can omit the wine and use vegetable broth if desired. Serve with crackers, bread, or crostini.

November 2022, HCU & You: Ask Methia

Dear Methia

The Simplified Diet: is this something that I can try?

I'm sure I'm not the only person with HCU to tell you this, but tracking and counting the amount of protein in everything I eat is EXHAUSTING. I know how important it is, and it really does help me keep my levels under good control. I'll admit, though, that some days I will forget to log something because, "Life Happens!" A friend of mine who goes to another metabolic clinic recently mentioned that she was trying something called the Simplified Diet. I'm confused as to what it really is, but I get the impression that she doesn't have to count everything she eats? She has PKU – would I be allowed to try something like this, too?

Sincerely,
Burned Out

Dear Burned out,

If I had a dollar for everyone who told me that they hated tracking their intake, I'd be able to pay someone else to write this column! It's absolutely an exhausting task. You're absolutely right, though, in that it's often necessary to track to make sure you are properly managing your HCU. That being said, the Simplified Diet is a relatively new concept being utilized by many metabolic clinics. Developed originally for people with PKU, it considers certain low protein fruits, vegetables and medical foods to be "free" or "uncounted." This doesn't mean that their protein allotment remains the same, though – clinics will typically adjust a person's protein (or phe) prescription to account for consumption of these free foods. In many studies, metabolic control remained unchanged when people stopped counting some of these "free" foods. I think it's perfectly reasonable to ask your clinic about taking a Simplified Diet approach to your HCU management. Here are some key factors to consider:

- **Good candidates have a slightly higher protein tolerance.**
  People who are already very restricted in natural protein/methionine may have difficulty with following a diet that will ultimately be even more restricted in foods that are not "free." Additionally, some "free" foods may not be considered negligible if your needs are already very low.
- **The Simplified Diet may actually "health-ify" your diet, too!**
  Since so many fruits and vegetables are uncounted, you will probably be more likely to reach for a free fruit or vegetable over that bag of potato chips in a pinch. It's also great to have a free food in your back pocket if you're hungry at the end of the day and have already reached your protein goals.
- **You may feel less anxious and more relaxed about your diet.**
  Even just knowing that some foods are ALWAYS an option, no matter where you are in terms of meeting your daily goals, can be very comforting.

Also, please remember that guidelines will vary from clinic to clinic.
Different clinics will have a different list of foods that are "free," may adjust your protein/methionine goals differently, and may even have different quantities of free foods that are permitted. Make sure you clarify with your clinic what these guidelines are, and never be afraid to ask questions!

Sincerely,
Methia

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

MEDICAL ASSISTANCE

What is the purpose of this program?

Having a rare disease is difficult. Adding in the complex care required to treat or manage that disease and figuring out how to pay for it makes a rare diagnosis even harder.

NORD's Classical HCU Medical Assistance Program offers eligible individuals diagnosed with Classical Homocystinuria financial support to pay for the low protein foods necessary in managing this HCU diagnosis.

NORD's HCU Medical Assistance Program opened thanks to a generous donation from the HCU Network America.

Who is eligible to apply?

This program is designed to help patients who:

- Have a diagnosis of Classical Homocystinuria.
- Are a United States citizen or U.S. resident of six (6) months or greater with evidence of residency such as a utility bill showing the patient's name and address.
- Meet the program's financial eligibility criteria.

What is the application process?

Patients may be referred to the program by their health care provider, their case managers, or they may self-refer.

A NORD Patient Services Representative will guide the applicant through the application process and verify eligibility for inclusion in the HCU Program.

Awards are based on meeting eligibility criteria, funding availability, and are made on a first-come, first serve basis.

NORD is Here for You

NORD, a 501(c)(3) organization, is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.

NORD was founded by families struggling to obtain access to treatments and whose advocacy for change led to the passage of the Orphan Drug Act in 1983. NORD assists eligible patients (those with medical and financial needs) in affording the treatments and medical services their healthcare professionals have prescribed.

Alone we are rare. Together we are strong.®

How do I get more information and apply?

Contact NORD's Classical HCU Medical Assistance Program

Monday-Thursday 8:30am – 7:00pm ET
Friday 8:30 am – 6:00pm ET

203-616-4327

203-635-4163

hcu@rarediseases.org

US MAIL to: NORD
Attention: HCU Program
55 Kenosia Avenue
Danbury, CT 06810

What assistance does NORD provide?

NORD's program can assist eligible individuals with the expense of purchasing low protein foods:

- The Classical HCU Medical Assistance Program assists eligible individuals with out-of-pocket costs to purchase low protein foods. Individuals approved for assistance in this program will be issued a PEX card. The PEX card is a prepaid expense card to be used for the purchase of low protein foods only.
- *Upon receipt of the card, the cardholder will contact NORD to request card activation. The card will be funded based on program award caps set for the program (this cap will be discussed with individuals upon enrollment in the program..*
  - *> It is necessary for the individual to submit receipts on a monthly basis evidencing card utilization for the purchase of low protein foods for the previous month.*
  - *> Funds will not be added to the card until the previous month's receipts have been received by NORD.*
  - *> The card may only be utilized for the purchase of low protein foods up to the monthly program limit.*

Once a patient is accepted into the assistance program(s) how long are they eligible?

Awards are issued for a calendar year.

Patients are encouraged to reapply annually if continued assistance is needed.

Program assistance is dependent on funding availability.

What happens if an applicant does not meet the criteria of the Electronic Income Verification?

The NORD Patient Services Representative will offer to e-mail, fax, or mail the brief program application and disclosure forms to the patient. The applicant may then complete the application, sign the disclosure form, provide the appropriate financial documentation to verify financial need, and return them via fax, email, or USPS mail.

How does NORD demonstrate compliance with regulations required of charities?

- NORD independently designs its patient assistance programs based on the needs of specific patient communities.
- No pharmaceutical company or donor controls or influences our programs.
- Our patient assistance decisions are based on consistently applied financial eligibility criteria and diagnosis only.
- Patients have their choice of health care provider, treatment and treatment location, and can make changes at any time.
- Patients' privacy and well-being are priorities at NORD. We do not share or provide patient names or data with donors, nor do we disclose or identify donors to patients. Patients are able to make the choices that are best for them because NORD's assistance covers all FDA-approved products available for a diagnosis. Our programs also help with more than medication: patients can use their funds to pay for other physician prescribed services related to their diagnosis, such as laboratory and diagnostic testing, physical and occupational therapy, durable medical and adaptive equipment, and travel to medical appointments.

NORD®

rarediseases.org

HCU AWARENESS MONTH RECAP

Social Media Reach

Social Media is a powerful tool for raising awareness!

Not only does HCU Awareness Month bring attention to our organization and the disease, but it also draws attention to other issues related to homocystinuria. We see a lot of growth thanks to those who follow along with us and share!

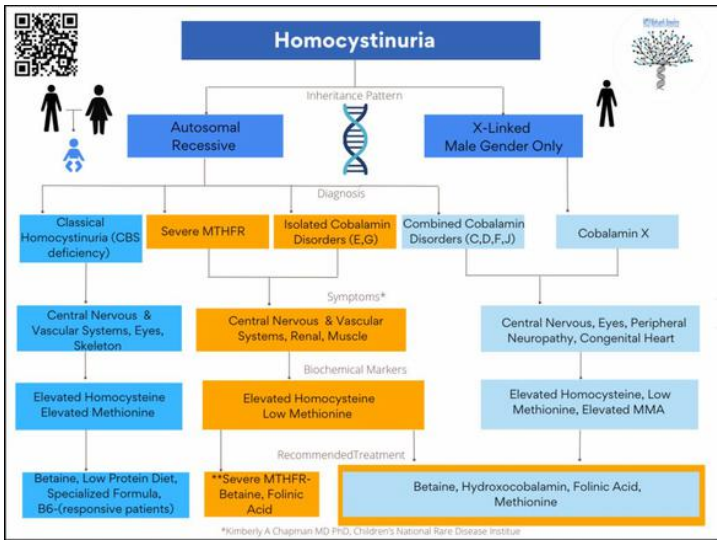
During the month of October, we gained:

- **20** new Facebook followers & grew our reach by **80**%
- **38** new Instagram followers & grew our reach by **180**%
- **21** new Twitter followers & grew our reach by **283**%
- **64** new LinkedIn followers & grew our reach by **47**%

With 17 shares, & 266 post engagements,
our HCU Awareness 2022 Day 1 fact had the biggest reach.

#HCUAwareness 2022: #HCUFact1 #Homocystinuria is an elevation of the amino acid, #homocysteine in our urine or blood. High homocysteine can be caused by #Cystathionine-Beta-Synthase (relates to B6), #Cobalamin disorders (relates to B12), remethylation disorders, such as #MTHFR (relates to Folate).

To download the Homocystinurias Flowchart graphic, visit https://bit.ly/HCUFlowChart



Thank you to everyone who participated in activities and shared during our 2022 HCU Awareness Month!

HCU AWARENESS MONTH RECAP

FABULOUS HCU AWARENESS FUNDRAISERS

Thank you for your support!

What is Amazon Smile?
Amazon Smile is a simple and automatic way for you to support HCU Network America every time you shop, at no cost to you. When you shop, you'll find the exact same low prices, vast selection and convenient shopping experience all with the added bonus that Amazon will donate a portion of the purchase price to us.

How do I set it up?
Simply, go to smile.amazon.com, the first time you enter the site it will ask you to designate an organization. Type in HCU Network America and select us from the list. It is important to note that in order for the donations to go to HCU Network America, you MUST check out from this url every time - see best practices below for some pointers on how to do this.

What if I'm already set up and would like to switch to HCU Network America?

1. From your desktop, simply select "Your Account" from the navigation at the top of any page
2. Then select the option to "Change your Charity". From your mobile browser, select "Change your Charity" from the options at the bottom of the page.
3. Type HCU Network America in the search bar and search for the charity.
4. Select HCU Network America charity to update your account
   If you are still having trouble, visit https://hcunetworkamerica.org/amazon-smile/ for the steps with images of how to.

Best practices for using Amazon Smile on a desktop
Now that your account is set up to use Amazon Smile, it is important to note that Amazon only makes donations to HCU Network America when you checkout from your cart from this smile.amazon.com. This is the only way HCU Network America gets any donations from Amazon Smile.

Shopping from your phone? Android and iPhone users, rejoice – you can now shop Smile.Amazon from the app – check out the instructions here- https://www.amazon.com/b?ie=UTF8&node=15576745011

What is Giving Tuesday?
GivingTuesday is a global generosity movement unleashing the power of radical generosity. GivingTuesday was created in 2012 as a simple idea: a day that encourages people to do good. Since then, it has grown into a year-round global movement that inspires hundreds of millions of people to give, collaborate, and celebrate generosity. This year, GivingTuesday will be **November 29**.

How do I get involved?

We are asking you to assist us in reaching our $10,000 GivingTuesday fundraising goal.

To get started, set up your own GivingTuesday fundraiser on GoFundMe.com, Facebook, or Instagram! Setting up your fundraiser on these platforms is simple and HCU Network America receives 100% of the donations!

- Start to reach out to your friends and family in advance and get them to pledge a donation first thing on November 29th!
- Let them know that by giving to HCU Network America, they are supporting programs and resources that directly benefit the patients and caregivers of our community.
- Remind them that all donations are tax-deductible AND will be matched by 3 anonymous donors (up to $30,000!)

How do I set up a GoFundMe fundraiser?

It's easy! Select 'charity' and type in HCU Network America. From there, just follow the prompts! GoFundMe is great because you can link in a video if you'd like. Consider a quick video telling how our programs have made a positive impact on you, along with your ask. Or, you can always post an image along with text.

To see a sample fundraiser from HCU Awareness month on GoFundMe, visit: https://www.gofundme.com/f/elliotts-hcu-awareness-2022-fundraiser

GIVING TUESDAY

OUR MATCHING GIFT IS BACK - AND BIGGER THAN EVER!!!

That's right, you heard us right! Thanks to two anonymous donors, **any funds** you help raise from October through December 31, 2022, will be matched **up to $30,000!**

We are asking every patient and family to help us raise funds for homocystinuria. During the winter holiday's warm hearts and generosity can be felt near and wide. During this time, we ask that you share our appeal letter with your colleagues, friends and family.

See our appeal letter on next two pages, or you can print it from [here]

EMPLOYER MATCHING GIFT PROGRAM

Did you submit for your Employers Corporate Matching Gifts Program?

What are Matching Gifts?
Corporate matching gifts are a type of gift giving in which companies financially match donations that their employees make to nonprofit organizations. When an employee makes a donation, they need to request the matching gift from the employer, who then makes their own donation. Companies usually match at a 1:1 ratio, but some will match 1:2 or 2:1.

Why are Corporate Matching Gifts Valuable?
Corporate matching gifts are valuable because they are free money for your nonprofit of choice, HCU Network America! Your donation has double the power without you having to give double the amount.

Does HCU Network America Really Benefit?
If corporate matching gifts weren't valuable, we wouldn't be sending this letter. In 2020 we received approximately $45,000 in corporate matching gifts!

How Do I Find out if my Employer has a Corporate Matching Gifts Program?
Your company website or websites such as doublethedonation.com make it easy for you to search and see if your employer takes part in corporate matching gifts. If you can't find it online, please consult with your employee handbook, HR rep or manager to find out.

How Do I Request my Donation is Matched by my Employer?

1. The donor completes their donation
2. The donor submits matching gift request
3. Company reviews donation and nonprofit eligibility and reaches out to nonprofit
4. Nonprofit verifies the donation was made
5. If eligible, the nonprofit will receive the matching gifts request!

Top Matching Gift Companies

Company	Match Ratio
General Electric	1:1
Gap Corporation	1:1
ExxonMobil	3:1
Johnson & Johnson	2:1
Microsoft	1:1
Pfizer	1:1
Coca-Cola	2:1
And many more!	

Did you know some companies match retired employees donations?

15 S. Mallory Ave
Batavia, IL 60510

(630) 360-2087
info@hcunetworkamerica.org
http://www.hcunetworkamerica.org
Tax ID Number: 81-3646006

Dear, First Name

The past two years have been a struggle for many. The year of 2022 feels oddly like a new beginning, though we recognize that challenges remain that affect your lives. We sincerely hope you have been able to successfully manage these challenges.

These times are extra demanding for people with unique medical needs and special diets, who spend time at clinics and waiting for life giving medical supplies. People with HCU must balance all the variables in their lives that affect their health and that can sometimes seem overwhelming.

HCU Network America is here to help this special group of people with the support and resources they need to navigate daily life. We are proud of the reach we have and the way these communities have knit themselves together. But we can't do it without your help! As a 501(c)(3), we need your donations, which are tax deductible, to continue with our mission and meet our goals. What you do to help us can make a huge difference in the lives of all HCU patients and their families.

We would like to share a story of a family's journey of diagnosis.

> *Our daughter was born in the summer of 2020. She was our second daughter, and we were thrilled to have a new addition to our family. We received a call after only one week that our daughter's newborn screening was abnormal. We were assured that it was most likely nothing, but we needed to do another screening to be sure. The second screening was abnormal as well. This time, we needed to go to our nearest metabolic clinic 4 hours away to test her levels, do a genetic test, and meet with the geneticist to learn about Homocystinuria and available treatments. The genetic test later confirmed a diagnosis of Homocystinuria due to severe MTHFR deficiency. Her initial Homocysteine level was 295 (normal levels are below 15), and her Methionine was 3 (normal levels are over 20). Although our daughter's condition was caught early, she didn't begin treatment until three weeks of age. During those first three weeks, our daughter didn't gain any weight and she slept more than normal. I initially thought she was a great baby who loved to sleep, but I later realized it was due to her condition. Within a few days of starting treatment, she started becoming more alert. At that time, I noticed it was difficult for her to focus her eyes. When she tried to look at me, her eyes would uncontrollably roll up. We brought her to the local ER, and the doctor believed that our daughter was having seizures. The metabolic specialists arranged for our daughter to be flown to them so that she could be monitored through the weekend. Thankfully, all tests came back normal, and for the first time since she was born 3 ½ weeks earlier, she was actually able to look and stare at me. After only one week of treatment, she gained more than a pound. For the first 7 months, she was consistently behind developmentally by 1 month. It was as if she completely "lost" that time before treatment was started. It was also months before her head size ever made it onto the growth chart. Our daughter is now two, and she is doing better than we ever expected. Besides a speech delay, she is developing like any other two-year-old. She is incredibly smart and the happiest child. Newborn screening and early treatment saved her life!*

Since HCU Network America was available, this family was able to get the support they needed at one of the most devastating times in their lives. In June of 2016, HCU Network America was incorporated; bringing hope to families living with HCU that they had advocates to help them get the latest and best advice from the medical community. HCU Network America also financially supports research that can help find new treatments. Since 2016 we have communicated with metabolic clinics all over the country

to reach out to new patients and provide them with toolkits which are filled with helpful tips and guidelines for living with HCU.

Here are some of the highlights that your donations helped with:

- Awarded four research grants for improved therapies and one for better newborn screening.
- Held our third patient/family-expert conference
- Launched a low-protein foods assistance fund
- Provided a consultant with experience in medical insurance to fight for coverage for medications and food at no cost to HCU patients
- Supported community meetups and monthly community newsletter
- Enrolled 42 patients in our Homocystinuria Data Collection program and 23 in our biobank repository
- Advocated for improved newborn screening which led to several states adopting changes

Our goal this year is to raise $60,000. Thanks to an anonymous donor, any funds you donate up to December 31, 2022 will be matched up to $30,000. Please consider a donation now to HCU Network America. If you personally know a patient with HCU, you can donate in their honor. Ask your families and friends to help. We need your help and appreciate any donation. In addition, if your employer matches charitable donations, they will match those too!

Take a minute to look at our website to see what we are up to and meet some our "heroes ": https://hcunetworkamerica.org. You can donate through our website or by mail.

Thank you for all you do to help us. We will all get through this year and look forward to 2023!

Thank you,

Danae' Bartke

Danae' Bartke
HCU Network America, Executive Director

Donor levels:

- **Leadership Circle** - $5,000 or more
  Donor's name, HCU Patient's name and photo on homepage of website, along with certificate donation
- **HCU Champion** - $1,000 or more
  Donor's name, HCU Patient's name and photo on HCUNA donation page along with donation certificate
- **HCU Supporter** - $500 or more
  Donor's name, HCU patient's name on website, along with a donation certificate
- **HCU Ally's** - $100 or more
  Donor's name and HCU patient's name listed on website

4 ways to donate:

1. Use the enclosed slip and envelope
2. Go to https://hcunetworkamerica.org/donate
3. Text HCU2021 to 44-321
4. Scan the QR Code

INDUSTRY NEWS

Aeglea BioTherapeutics Provides Clinical Progress and Regulatory Update for Homocystinuria Program

AUSTIN, Texas, Oct. 4, 2022 /PRNewswire/ -- Aeglea BioTherapeutics, Inc. (NASDAQ:AGLE), a clinical-stage biotechnology company developing a new generation of human enzyme therapeutics as innovative solutions for rare metabolic diseases, today announced that dosing in the third cohort of the Phase 1/2 clinical trial of pegtarviliase for the treatment of Classical Homocystinuria is underway.

Additionally, the company received a letter from the U.S. Food and Drug Administration (FDA) responding to a recently submitted protocol amendment for the Phase 1/2 clinical trial of pegtarviliase for the treatment of Classical Homocystinuria, saying that additional information is needed before the trial can be expanded to adolescents.

To read the full press release, visit https://bit.ly/3ChO30M

Travere Therapeutics Reports Third Quarter 2022 Financial Results and Corporate Updates

SAN DIEGO, Oct. 27, 2022 (GLOBE NEWSWIRE) -- Travere Therapeutics, Inc. (NASDAQ: TVTX) today reported its third quarter 2022 financial results and provided a corporate update.

A Breakthrough Therapy Designation was granted to the pegtibatinase development program for classical homocystinuria (HCU).

To read the full press release, visit https://bit.ly/3U5MdY9

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

Say "Cheese" for Oral Health!

From short roots to crowding of teeth patients with various types of homocystinuria experience a wide range of eye issues. While some of these dental issues are better documented, other aspects aren't.

You know the  **drill, complete the Oral Health Survey**

homocystinuria.rare-x.org

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

NEWBORN SCREENING

Liz Carter, mom of 6-year-old Elliott, is a Newborn Screening Ambassador!

Elliott was missed at Newborn Screening and was diagnosed at age 2 1/2 with Classical HCU after suffering serious health complications. By sharing Elliott's Newborn Screening story, Liz hopes to raise awareness of the gaps and limitations within the Newborn Screening system so that all babies - no matter where they live, can be identified at birth, giving them the best chance for a healthy start to life!

To read more about Liz's ambassador journey, and to meet the other ambassadors, visit https://bit.ly/3DR9Leh

Be sure to check out these ***Navigate Newborn Screening*** Ambassador videos, where Liz shares more about Elliott's story and her experiences in the Ambassador program:

NBS in one word | Meet the ambassadors | Community Leadership

NEWBORN SCREENING

Meet the Ambassador, August 2022

I am an NBS AMBASSADOR

A Warm Welcome from Liz Carter

South Carolina | carterliz84@gmail.com

Hi, I'm Liz! I live with my husband and 2 boys in Lexington, South Carolina. I am a mom, a former Spanish teacher of 14 years and now, an advocate for those living with Rare Diseases as well as a Newborn Screening Ambassador! While my journey has not always been an easy one, it's led me to exactly where my passion lies and where I feel I'm supposed to be. I'm happiest when I'm spending time with my family and our dog, and when I can use my experiences and my voice to help others and to create positive change. Becoming an Ambassador for Newborn Screening has been an opportunity for me to both walk alongside other families as they navigate their own Newborn Screening journeys, as well as provide an avenue to learn more about a system that has been proven incredibly effective for many, but through the sharing my family's own experiences, I am able to advocate for improvements.

In 2013, I gave birth to our first son, Grayson, who passed his newborn screening tests with flying colors. 3 years later, our family welcomed baby Elliott, who from the moment he came in to the world, lit up our lives. Just like his brother, Elliott aced his newborn screening tests! (Or so, we were told in the hospital after his arrival.). We took Elliott home, and just like that, our family was complete. In 2018, our family took a trip to the beach. When we returned home, Elliott (age 2 1/2 at the time) became very sick. We thought he had a virus and watched and waited for it to pass. It didn't.

We ended up in the Children's ICU, where Elliott was placed in to a medically induced coma. We were told that he was having seizures and that they needed to stop the seizure activity to give his little brain a chance to rest. We soon found out that the seizures were a result of extensive clotting throughout Elliott's brain. But WHY? Doctors told us that we may never find out the cause. They told us to brace ourselves; that we could lose Elliott to this. Grasping at straws and racing against the clock, doctors dug deep to determine what was going on. 14 days later, we got the news. "A recent blood test shows a very high level of blood Homocysteine, and we think Elliott has something called Homocystinuria. It's a rare, genetic metabolic disorder. The good news is, it can be managed with a Low protein diet, medical formula and medication." ....And that's where our 'new normal' began. I share our story as often as possible, with anyone that I can. Becoming a NBS Ambassador has given me the opportunity to support other parents that maybe have had a similar experience with a missed diagnosis at screening. It's also allowed me to lean on other moms/caregivers when I need the support myself! I can't say enough about the positive impact that joining the Ambassador Program has had on my life, and hopefully has allowed me to leave a positive impact in the lives of others.

Navigate Newborn Screening

MEET OUR NEW COMMITTEE MEMBER!

Danielle is a patient with Classical Homocystinuria diagnosed through the Newborn Screening Program in New York City. She resides in Winter Park, FL with her husband Irving, 3 sons and 3 dogs. Thankfully, having HCU is all she knows and has experienced minimal negatives and multitudes of positives. Danielle comes from a large family that serves as an amazing support system which continuously feeds her passion of servant leadership amongst the HCU community. Danielle is excited to lend her experience as a patient, traveler, foodie, and learning and development professional within the “big 5” health care companies to promote that living with a rare disease while simultaneously obtaining success in your passions is possible.

EVENT ANNOUCEMENT

JOIN US
FOR A VIRTUAL COOKING CLASS!

REGISTER HERE:: HTTPS://WWW.EVENTBRITE.COM/E/456507144157

MEET OUR NEW COMMITTEE MEMBER!

Brandon lives in the high desert of Southwest Wyoming in the small town of Green River with his wife Shandra and two children, Madyson and Mason. Their story in the HCU world began when Mason's newborn blood screening picked something up and he was sent to Children's Hospital of Colorado's NICU where he was diagnosed with Cbl C. Brandon is an avid mountain biker and enjoys camping, fishing, and helping with Mason's Cub Scout pack.

NEW RESOURCE

Recomendaciones para el control de los trastornos de la cobalamina

¡Ahora disponible en español!

Visitas a la clínica genética o metabólica

Fuentes: Huemer et al. 2016 https://doi.org/10.1...
Sloan et al. 2018 https://www.ncbi...

Primer año de vida 1-2 veces/mes según sea necesario hasta que el control metabólico y el crecimiento se estabilicen

Niño pequeño en edad escolar 2-4 veces/año

Adolescente adulto 1-2 veces/año

Monitorear en cada visita a la clínica

HCU NETWORK AMERICA

Área a vigilar: Valores recomendados

...ol/L <12 años <60 umol/L Adultos <100 umol/L Enfermo <100 umol/ L

...<10 umol/L

...ia
...ura: Crecimiento deficiente
Peso: Desnutrición, poco aumento de peso
Circunferencia de la cabeza: Microcefalia

Dieta:
Diario de comidas: Alimentación deficiente, Cumplir con los objetivos calóricos y proteicos

Monitorear anualmente

Área a vigilar: Problemas asociados

Exámenes de laboratorio nutricionales/metabólicos

Vitamina B12
Aminoácidos plasmáticos: Seguimiento de la metionina para ajustar la betaína y la OHCBL
Recuento sanguíneo completo: Citopenia, anemia megaloblástica
Prealbúmina: Desnutrición, función hepática
Albúmina/Proteína total: Desnutrición, función hepática

Ácidos orgánicos en orina
ALT/AST
Electrolitos
Función renal
Carnitina plasmática, total/libre

Oftalmología

Examen de la vista: Maculopatía, Retinopatía, Detección y gestión del estrabismo
(fotografía de la retina, tomografía de coherencia óptica, ERG (cada 6 meses los primeros 2 años):
Evaluación oftalmológica: Remisión a recursos/servicios de baja visión para personas con discapacidad visual

Riñón

Análisis de sangre: Creatinina, nitrógeno ureico en sangre, cistatina C: Enfermedad renal, microangiopatía trombótica, síndrome urémico hemolítico
Análisis de orina: Análisis de orina

Neurología y desarrollo neurológico/neuropsicológico

Examen clínico: Hipotonía, convulsiones, neuropatía periférica, movimientos anormales, hitos del desarrollo,
Necesidad de servicios, IEP

Cardiovascular

Revisión de los factores de riesgo cardiovascular: Complicaciones tromboembólicas
Ecocardiograma: No compactación del ventrículo izquierdo, otras cardiopatías congénitas Presión arterial: Hipertensión

Monitorear según sea necesario

Área a vigilar: Problemas asociados

Laboratorios nutricionales/metabólicos

Ferritina/Hierro: Evaluación nutricional periódica
25-hidroxivitamina D
Ácidos grasos esenciales

Neurodesarrollo/Neuropsicología

Pruebas de CI: Deterioro cognitivo, evaluación del autismo

Psicología

Psicología clínica o evaluación psiquiátrica:
Trastornos mentales y del comportamiento

Medicina de rehabilitación

Medicina física y rehabilitación, fisioterapia, terapia ocupacional y logopedia

Neurología

Resonancia magnética del cerebro y la columna vertebral: Hidrocefalia, convulsiones, trastornos del movimiento. Degeneración combinada subaguda de la médula espinal
Electroencefalograma (EEG): Convulsiones
Estudios de electromiografía/conducción nerviosa: Neuropatía

Salud del hígado

Ecografía abdominal: Hígado graso
Cada 3-5 años a partir de la adolescencia
A menos que esté clínicamente indicado más temprano

Salud ósea

Imágenes DEXA de diagnóstico Cada 5 años a partir de la adolescencia, salvo que esté clínicamente indicado antes: Salud ósea

Download the Spanish Cobalamin Monitoring recommendations: https://bit.ly/MonitorCblSP

EVENT ANNOUCEMENT

Save The Date & Register TODAY!

MSRGN's Genetics Summit 2022:

Momentum in the Mountains

A Virtual Event
to be held on

November 9-10, 2022

Starting at:
8:30 am PT, 9:30 am MT, 10:30 am CT, 11:30 am ET

and ending at:
2 pm PT, 3 pm MT, 4 pm CT, 5 pm ET
(with breaks and lunch)

A virtual summit to educate, engage, and connect families, providers, and public health professionals around contemporary topics in genetics.

Register here

EVENT ANNOUCEMENT

Homocysteinemias online course

November 7-18, 2022

- The course targets primarily (but not exclusively) advanced practice providers and clinicians who:
  - have a science background
  - have had no/limited formal training in genetic metabolic disease
  - are interested, or are involved, in the diagnosis/management of patients with inborn errors of metabolism

- Free of charge for physicians & trainees, nurse/nurse practitioners, physician associates, genetic counselors, dietitians; $100 corporate applicants

Learn more and sign up here: https://bit.ly/3UPLfk7

Approved for 7.0 AMA PRA Category. 1 Credits™/ 7.0 AAPA Category 1 CME credits/ 7.0 ANCC Contact Hours/ 7.0 Credits for Dieticians

VMP, llc

Expanding Genetic Horizons

Participate in a Homocystinuria (HCU) Research Study

This study is looking at the safety of an investigational drug, pegtarviliase, and how well it is tolerated in patients with Homocystinuria (HCU)

About The Study

- Patients will participate across the globe
- Your participation will last approximately 14 weeks to include a screening period, 4 weeks of treatment, and a follow-up period
- You and your healthcare provider will know that you are taking the study drug (Open Label)
- Some study visits may be done at your home
- Study-related expenses (travel service, reimbursement for loss of earnings, and other study-related expenses) will be provided

Interested in Participating?

Please contact Kellyn Pollard and Juana Luevano at:

GeneticsResearch@utsouthwestern.edu

at The University of Texas Southwestern Medical Center in Dallas, TX

Why Participate?

- Help advance HCU research and therapies
- Help scientists understand how pegtarviliase works*
- You will be receiving pegtarviliase, a potential treatment before it is widely available

Who Can Participate?

- 18 years of age and older
- Diagnosis of HCU due to Cystathionine β-Synthase (CBS) deficiency
- For additional eligibility criteria, please scan the QR code below, or visit: https://bit.ly/3CX7XAd

*This study may be of no benefit to you. Taking part in this study may or may not improve your health.

aeglea.com

aeglea

NOW ENROLLING: A Natural History Study on Classical Homocystinuria

Travere Therapeutics is enrolling children and adults with classical homocystinuria (HCU) in a Natural History Study. The goal is to learn more about classical HCU and course of the disease. Information gained from this study may help to improve understanding of HCU and help other patients, families, healthcare providers, and researchers to design new clinical research studies and therapies. No investigational medicine will be given to patients.

Approximately 150 patients will participate at sites in the US, Europe, and other countries around the world. The study will include three key stages (screening, enrollment, and observational follow-up) and will last approximately 6.5 years.

You (or your child) may be eligible to participate in the Natural History Study if you:

- **Have been diagnosed with HCU**
- **Are 5–65 years of age**

You (or your child) will need to meet all other study criteria to take part in the Natural History Study.

For additional information about the Natural History Study, please go to: https://www.clinicaltrials.gov/ct2/show/NCT02998710

You may be able to receive payment for time and travel when you participate in this study. Talk with your doctor and family members about joining the Natural History Study. Sites are open and currently enrolling patients.

If you have any questions, please email:

HCUConnect@labcorp.com

Visit www.hcuconnection.com for more information

MA-PE-22-0004. March 2022.

TRAVERE THERAPEUTICS

Live better, together!

Contact Register

What is the contact register?

The contact register is a secured private survey that allows you to share information on you or your family member with HCU with us. This includes where you are from, your relationship to homocystinuria, the patient's birthdate, gender, their exact diagnosis (e.g. CBS, cobalamin, or MTHFR), how they were diagnosed, and if the patient was diagnosed through newborn screening. This information is kept confidential and will not be shared unless you give us permission. By registering, you will be able to identify other patients in your state and request their contact information. You will also be able to access information posted over time that can only be shared with the patient community. (For example, we may have webinars that the expert presenter does not want to be publicly available, but is willing to share with the HCU community.)

What will this information be used for?

HCU Network America strives to inform patients and families with resources, create connections, and support advancement of diagnosis and treatment of HCU and related disorders. The information you provide helps us succeed in our mission - plan events, develop resources and educational tools, and ensure everything is being done to support timely and accurate diagnosis from birth. It also allows us to have informed conversations with doctors, pharmaceutical companies, and law makers. Your information helps us understand the landscape better so we can better advocate for you!

How do I participate?

The contact register form takes approximately 3-5 minutes to complete. You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to: https://bit.ly/3OJuF1W