The HCU Herald

Featuring...

HCU Hero
Victor from Arizona

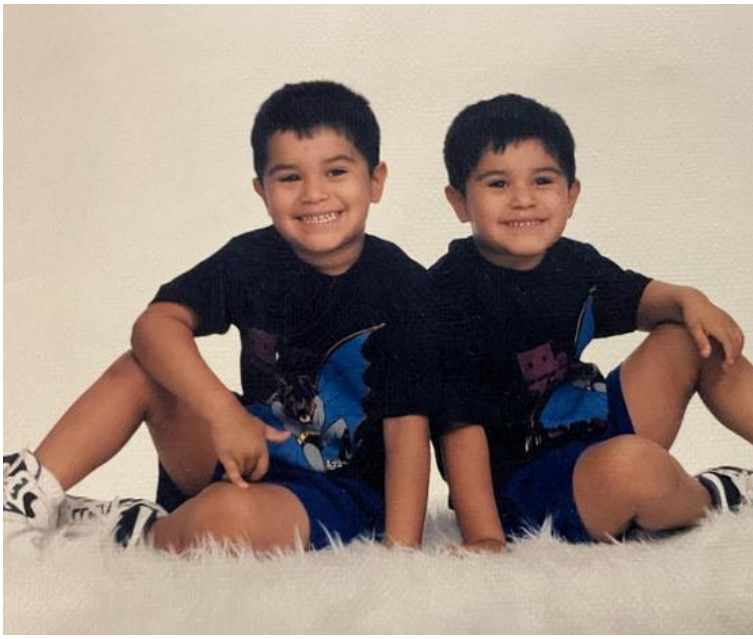
July 2023

HCU NETWORK AMERICA

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

HCU HERO: VICTOR FROM ARIZONA

In 1989 my twin brother and I were diagnosed with classical HCU as newborns. My parents told me they got a call from the hospital informing them that my newborn screening test was abnormal. They were concerned because they had never heard of this rare disorder before and didn't know what to expect. I was lucky enough that my parents started my HCU treatment and medications, along with food restrictions, right away when they got the information that they needed from the nutritionist.



Victor (*right*) & his twin brother Vicente (*left*)

Growing up with HCU was difficult, especially in elementary school. My mom would make my lunch every day until I was old enough to pack my own lunches. I missed out on a lot of pizza and ice cream parties. It wasn't until about 6th grade when I was able to make my own choices within the school cafeteria, and by that time I was pretty knowledgeable as to my diet and what I could and could not eat. High school was easy as well because I would just buy food from the café or vending machine to get me by until I got home from school. I enjoyed eating french fries and chips, and these were my go-to choices when I was younger in school (not the healthiest choices, but it worked for me!).

I would explain my condition to my teachers and friends by saying that my body can't break down high amounts of protein, so I'm on a low protein diet and if I eat too much of it, I can have a bunch of health problems as I get older. I eventually just started telling everyone that I was vegan, because it was becoming more popular and easier for people to understand, and if they kept prying, I would tell them I had HCU and that's why I was eating mostly vegan.

Formula and medication weren't too hard to manage, because my parents always stayed on me and my twin brother take it, every day. I used to hate drinking the formula as a kid, even with a bunch of orange tang to mask the flavor! As I got older, I developed a different pallet and can drink the formula with no flavorings. It does not taste good, but I'm used to the taste. After 34 years, you just deal with it!

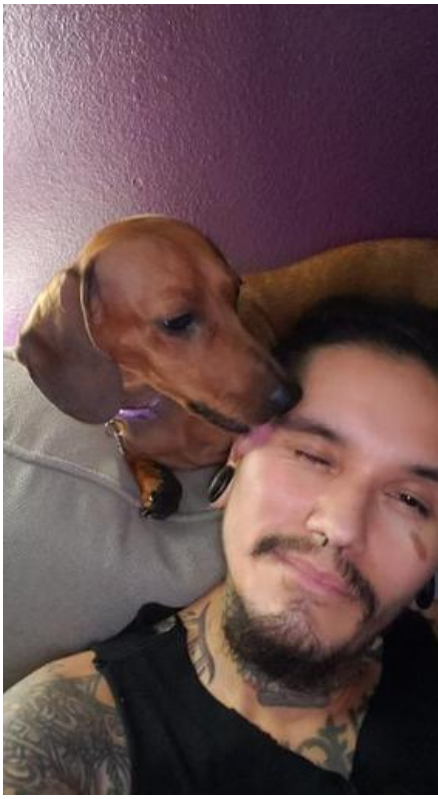
Growing up I enjoyed skateboarding, riding my bike, being outside and playing hide and seek or tag with my neighborhood friends. Even a game of baseball or tackle football was fun - we liked rough housing as kids. I loved playing Nintendo and PC games and I still enjoy gaming, especially playing RuneScape. It's been 20 years now of playing that game off and on.

As an adult, my disorder has never really affected my family. My family has always been very accommodating of my diet and at family functions, they've always set aside a special meal for me and my brother to eat.

My biggest challenge as an adult is remembering to take or refill my medications. My loving girlfriend helps a lot now by reminding me to take my medication or order more, so that I stay on track with everything. To help me keep track of medications, I will often write things on a calendar or set alarms on my phone. With time, it's getting easier to manage everything.

Another challenge that I've faced as an adult was having lung surgery, and the recovery that came with it. I've been dealing with Valley Fever for over 10 years now. Valley Fever is an infection that's caused by a fungus that grows in the soil in the Southwestern US and it attacks the lungs. There is no cure for it, just a control medication. When I was 22,

a lung doctor said I could stop taking the medication and my body would fight on its own and I shouldn't get sick again. However, at 29, the infection came back and had eaten away at my lungs. It put me in the hospital, and I almost died. Doctors were not sure if my HCU had anything to do with my immune system not being strong enough to fight it off or not. After the surgery, my lung also took over 3 months to heal, and because of my HCU, they were concerned about the potential for blood clots. But here I am today, 5 years later, and thankfully there have been no further complications, and my condition is stable. That was a very difficult time, but it made me stronger mentally and physically, and now I'm back in action riding my bike and going to Muay Thai, a style of boxing that I enjoy, 3 times a week.



Being on a low protein diet is another challenge that having HCU presents. Traveling is kind of hard, but I've learned to just research local vegan dishes whenever I plan to travel, and it works out. Speaking of the diet, I'm not always the best at it. But, for the most part, I still drink my prescribed formula and eat a vegan diet. I always check nutrition labels when available and I stay away from all meats, fish, and animal products. This has all helped me be able to manage my condition through diet really well over the years. I shop at Sprouts and Whole Foods often - you can really find a wide range of products that are vegan and low protein! It's all about reading the nutrition labels. And I still love eating the Cambrooke pastas!

> **You can really find a wide range of products that are vegan and low protein! It's all about reading the nutrition labels.**

As a working adult, my HCU has been easy for me to manage. I'm an HVAC-R technician, so I mostly work alone. I'll pack a lunch or stop and grab something at a local fast-food place, grocery store or gas station. My close co-workers know about my condition, so they'll ask me where I want to eat if we meet up for lunch because they understand my dietary restrictions. I like to keep my friendship circle small because I've always kept to myself - I can count on two hands my friends who know about my condition. The people who just know me or I consider my acquaintance just think I'm vegan.

My advice for anyone living with classical HCU is to drink your formula, take your medications, and read nutrition labels! I can't speak for everyone, but a vegan diet has been working well for me. I know everyone's body works differently, so be safe when experimenting with what diet works best for you. And don't be scared to live your life to the fullest! Go out in the world and try new foods that work within your diet or try a new activity! I've always lived my life saying you only live once, so I'm going to make the most out of it. (Can you tell that I've always been the wild twin?).

My hope for my future is just to live a healthy life and take care of my newborn son, Dazai. I'm hopeful that one day I'll be able to take a medication that will allow me to eat whatever I want without restrictions. I love food and traveling; the world is big, and I've seen so much food I would love to try one day! But, until then, I hope HCU gets more attention and support to find a cure or even something to make it okay to intake more protein.

To read Victor's story on our website, visit
https://hcunetworkamerica.org/victor-arizona/

Will YOU be our next HCU 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

JOIN OUR FUNDRAISING TEAM

We are looking for new community members to join our fundraising team!

Help create, organize and support new and existing fundraising ideas.

These vital funds help support our outreach, programs and research!

Email us:
dbartke@hcunetworkamerica.org

UPCOMING EVENTS

Register now for our EL-PFDD meeting!

What is an EL-PFDD meeting & what does it mean for our community?

Externally led patient-focused drug development (EL-PFDD) meetings bring together patients and care partners, FDA representatives, pharmaceutical companies, doctors who are experts in the particular disease, and other stakeholders.

For the meeting on Classical HCU, the goal is to hear from patients on what it's like to live with the condition so that we can better understand the patient experience. This information can help the FDA to make informed decisions on approvals of potential medicines for Classical HCU, and pharmaceutical companies to design clinical trials that are meaningful for patients.

COME JOIN US!

Community Meeting

September 28, 2023, at 4 pm ET

Come find out:

- Why is an EL-PFDD important to our community
- What to expect when attending the HCU EL-PFDD meeting

EL-PFDD Meeting

October 27, 2023, at 10 am to 3 pm ET

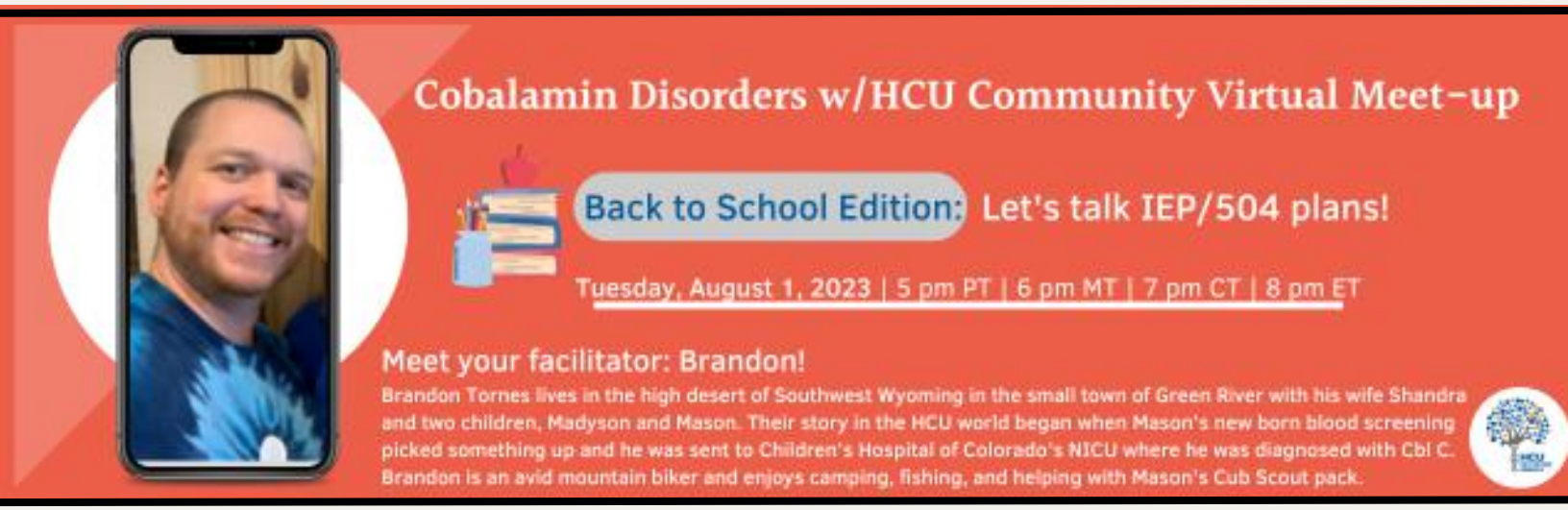
Register now

https://bit.ly/hcu-elpfdd

Click here to register for our EL-PFDD & our informational Community Meeting!

UPCOMING EVENTS

Register: https://hcunetworkamerica.org/virtual-meet-ups/



RARE ACROSS AMERICA

EVERY VOICE, IN EVERY DISTRICT, MATTERS

August 7-18, 2023

House meetings will be in-person only: Each advocate will have the opportunity to meet in-person with their Member of the U.S House of Representatives.

Senate meetings will be virtual only. Each advocate will have the opportunity to virtually meet with both of their U.S Senators.

EVERYLIFE FOUNDATION FOR RARE DISEASES

Register today!
Registration closes July 11.

Training webinars provided!

- **July 18th, 2:00 pm ET: Rare Across America General Training Webinar**
- **July 25th, 2:00 pm ET: Share Your Story with Policymakers Webinar**
- **July 27th, 2:00pm ET: Team Coordinator Training Webinar**

Click here to register!

Newborn Screening & HCU

International Neonatal Screening Day was observed on June 28, the birthday of Dr. Robert Guthrie, who pioneered newborn screening.

Newborn Screening Is a Lifesaver

Learn more about a new test to help babies with rare genetic disease get faster treatment.

Liz and Don Carter enjoyed their annual family summer vacation in Myrtle Beach, S.C., in 2018 with their two sons, ages 5 and 2. The boys thrilled to the magic and excitement of the ocean. They laughed, splashed in the water, and squished sand between their toes.

But the happy scene changed on the drive home. Elliott, the 2-year-old, became ill and started vomiting. He appeared to have a typical virus, but Liz Carter began to worry after a few days went by with no improvement. She took him to the children's emergency room, where the doctor seemed to chalk up her son's illness to a virus that would pass soon.

Liz, Grayson, Don, and Elliott Carter share precious time together.

(Photo courtesy of Liz Carter)

New Test Could Help Newborns with HCU Get Faster Treatment

Thanks to researchers at the Centers for Disease Control and Prevention (CDC), a new way to screen for HCU in newborns will soon be available.

The new test measures homocysteine levels directly. It can be done at the same time as other screening tests, and on the same blood spot sample from the baby. This will allow babies with HCU to receive care and treatment much faster, before any symptoms occur.

"Our new test makes homocysteine analysis easier, solving a two-decades-old problem, and hopefully making cases like Elliott's less common. The test increases the specificity for HCU screening," explains Austin Pickens, PhD, research and development team lead for CDC's Biochemical Mass Spectrometry Laboratory.

The CDC study on HCU screening was published in early 2023 in the American Association for Clinical Chemistry's *Clinical Chemistry* journal. "This is a significant step to greatly improve quality of life for infants with HCU," declares Konstantinos Petritis, PhD, chief of CDC's Biochemical Mass Spectrometry Laboratory.

Baby Elliott in intensive care

(Photo courtesy of Liz Carter)

Accurate testing means that care and treatment can start immediately, allowing newborns to avoid lifelong health problems or an early death.

Click here to read the full article!

Newborn Screening & HCU

Our Executive Director, Danae' Bartke, was featured in the June ***Spotlight on Newborn Screening*** edition of Rare Revolution Magazine!

Striving to do better

Homocystinuria (HCU) has been on the Recommended Uniform Screening Panel (RUSP) in the US for nearly 15 years, yet babies with the condition are still being missed at birth, risking serious health consequences. Danaé Bartke is the executive director of HCU Network America and has classical homocystinuria (HCU). She explains that while improvements in newborn screening processes can potentially help more babies to receive that vital early diagnosis, progress relies on each state updating their screening protocols; something her organisation is working tirelessly towards

Danaé's story

Although she is five years older than her brother Garrett, Danaé Bartke's story begins with his diagnosis in 1995, just before his fifth birthday. Danaé explains that when Garrett was born he missed every milestone. The family's paediatrician dismissed any concerns Danaé's mother had, saying that boys develop slower and he would get there eventually. Garrett was born sixth out of eight siblings and so they were also told that because he had other people to do things for him, he would always be less inclined to do anything for himself. But as Danaé explains, "In large families that's not the case, the younger you are in the line-up, the faster you progress. Number seven in our family walked at 10 months and number eight at just 8 months."

Just before Garrett turned five, the lens in his eye dislocated, which is one of the hallmarks of classical homocystinuria (HCU). His eye doctor referred him to a specialist, who suggested genetic testing was carried out. Within a month Garrett had been diagnosed with HCU. Following Garrett's diagnosis, the rest of the family were tested, and out of the eight children, Danaé was the only other sibling to test positive for HCU. She was 10 years old.

> "I'm incredibly thankful to be able to share this journey with my brother. And I'm really thankful that I was asymptomatic—any indications I did have were sensory quirks that could very easily have been written off as well. Despite Garrett still experiencing some delays from his late diagnosis, compared to other families we've been pretty lucky."

What is homocystinuria (HCU)?

Homocystinuria refers to a group of inherited disorders in which the body is unable to process certain building blocks of proteins (amino acids) properly. This leads to increased amounts of homocysteine and other amino acids in the blood and urine. The most common type of genetic homocystinuria, called CBS deficiency, is caused by the lack of an enzyme known as cystathionine beta-synthase (CBS). Homocystinuria can affect the eyes, skeleton, central nervous system and the blood clotting system.

https://rarediseases.org/gard-rare-disease/homocystinuria/

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CHARITY | ADVOCACY

Danaé pictured with her brother Garrett who is five years her junior. Garrett was diagnosed with homocystinuria (HCU) just before his fifth birthday

Treating HCU

There are two types of HCU: pyridoxine (vitamin B6) responsive and pyridoxine non-responsive. Pyridoxine therapy is used to reduce the levels of homocysteine and methionine in the body and is effective for approximately 50% of individuals.[1] For those who do not respond to this therapy, a diet that is low in protein and methionine is advised, for life. This restrictive diet can be hard to maintain for people who are not diagnosed until childhood, adolescence or adulthood, as their tastes have already evolved. Individuals on this diet also require supplemental metabolic foods, usually in the form of a formula, to provide them with other essential amino acids. Alongside the restricted diet, people may also be prescribed a medication to help lower the levels of homocysteine in the body.

A set of guidelines for the diagnosis and management of cystathionine beta-synthase (CBS) deficiency, the most common type of homocystinuria, was published in the Journal of Metabolic disorders in 2017. The recommendation is for those who are pyridoxine responsive to keep their homocysteine levels below 50, and for those who are pyridoxine non-responsive, below 100. It has been found that if patients maintain the recommended guidelines, the risk of blood clots and lens dislocation is significantly reduced. "We know if patients get diagnosed at birth and put on a treatment plan, then we mitigate the possibilities of the red flag-type symptoms. Some patients may still have basic myopia or less severe symptoms, but overall it's controlled."

The importance of early diagnosis

There are many consequences of a late diagnosis of HCU, which can worsen if left untreated. The effect on the eyes can be acute. Many people with HCU are short-sighted but, if untreated, their vision can deteriorate over time, often meaning a loss of independence. Gradual lens dislocation can also result in blindness. Garrett's lenses were eventually removed and Danaé experienced lens dislocation at college in her early twenties. "It was terrifying because I could see what was happening."

Strokes and blood clots are another serious risk from a late diagnosis. Sadly, blood clots have proved fatal for many young children with HCU. Patients are also at a higher risk of scoliosis and osteoporosis starting in early childhood.

Danaé says other less-documented symptoms are the effects on a person's mental health, along with behavioural disorders. "Patients can experience anxiety, depression, anger, aggression and obsessive-compulsive type behaviours."

Missed diagnoses

HCU was added to the Recommended Uniform Screening Panel (RUSP) in the US in 2009. The RUSP is a list of disorders that are recommended by the US secretary of health and human services for states to screen for. However, since its formation in 2016, the HCU Network America has frequently been contacted by patients whose HCU was not identified by newborn screening (NBS) but diagnosed several years later, due to the onset of symptoms.[2] Experts have estimated that 20% to 50% of pyridoxine non-responsive HCU patients (who are more severely affected) are missed by current NBS approaches.[3]

Why, when HCU is screened for at birth, are so many babies being missed? Danaé explains that there are a few contributing factors. Firstly, it is mandatory to carry out NBS within the first 24 to 48 hours in the US. To identify HCU in babies through NBS, methionine levels are tested. Methionine is not produced by the body but comes from the ingestion of food. When babies are tested at 24–48 hours, not much methionine has been introduced into their bodies, partly because breast milk is very low in methionine and because it hasn't had enough time to accumulate in their bodies.

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DID YOU KNOW?

Homocystinuria was added to the Recommended Newborn Screening Panel (RUSP) in 2009, but experts have estimated that **up to 20-50% of B6 non-responsive HCU patients are missed by current Newborn Screening approaches**.

Read the full article (on pgs 12-15) to learn more about HCU Network America's mission to improve Newborn Screening for HCU!

Thank you to **Rare Revolution Magazine** for helping to bring awareness to this important issue!

UPDATE

Now includes funding assistance for medical formula AND low protein foods!

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

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 and physician prescribed medical formulas necessary in managing this HCU diagnosis.

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

MEDICAL ASSISTANCE

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?

- The Classical HCU Medical Assistance Program assists eligible individuals with out-of-pocket costs to purchase low protein foods and physician prescribed medical formulas. 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 or physician prescribed medical formulas 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 or medical formulas 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.

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.

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NORD®

rarediseases.org

Cookie Butter Pie

Makes 1 pie= 16 small slices per pie | Serving size 1 slice = 1.3g protein and 61mg PHE

Ingredients:

- 4 TBSP Butter, softened
- 227 g Daiya Cream Cheeze, Plain, room temperature
- 1/2 c Creamy Speculoos Cookie Butter
- 1 tsp Vanilla Extract
- 170 g Cool Whip
- 1 crust(s) Graham Cracker Crust, 9-inch

Directions:

1. Prior to mixing make sure the butter and cream cheese are soft enough to cream together. In a mixer with a paddle attachment, cream together the butter and cream cheese until light and well blended. Add the cookie butter and vanilla and mix until creamy. Remove the mixing bowl and fold in the cool whip. Try not to over mix as the cool whip helps to make the pie light and fluffy.

2. Pour the filling into your graham cracker crust. Refrigerate for at least one hour to set. Serve and enjoy!

Notes:
You can prepare your own graham cracker crust using Brenda's graham cracker on HowMuchPhe recipe to lower protein or PHE.

July 2023, HCU & You: Ask Methia

Dear Methia,

I need some new flavors in my life!

I am bored with the foods I eat! I feel like I have been eating the same foods every day for my whole life! I am so tired of the same steamed veggies or low protein pasta and red sauce that I am often tempted to veer off diet (I don't though!). I read about adding spices or different flavors to foods, but I get so nervous about it having protein or knowing what goes with what. Can you help?!

Thanks!
Help Me Spice It Up

Dear Spice it Up,

Flavor fatigue is a very common thing. Most people get into a food rut, even those who do not have special diets. Spices can add so much flavor and help give variety, along with trying different cooking methods! Let me walk you through a few things that you can start implementing today!

Spices 101: Spices can take you on a journey around the world! You can add one spice or many, creating different flavor profiles and increasing the flavor to your liking. Spices are the seeds, leaves, roots, and sometimes flowers of a plant. They can be used whole or ground and come in fresh or dried.

What about adding protein? Ground spices are typically zero protein, which makes them great for a low-protein diet! To note, it's important to be aware when using spice mixes or whole seeds in large amounts, as the mix may include additional ingredients that may have protein. Best to always glance at the label before using, to be safe!

How to use? There really is no wrongs when it comes to spices! Use them before, during or after cooking. The more you use, the stronger the flavor. You can play with what you like and experiment! There are great resources online to find common uses and combinations for spices. For example, basil and tomatoes or everything but the bagel seasoning and avocado! Cinnamon and honey are a great way to bring out the sweetness of winter squash or sweet potato. I encourage you to pick up a couple of spice mixes the next time you are at the grocery and experiment with different foods. You can even mix them with an oil of your choice to make a quick vinaigrette to top salad, pasta, or roasted veggies!

Bonus Tip: Spices can add so much, but so can cooking your foods in a different way! Always steaming from a frozen bag? Try roasting in the oven or using an air fryer to bring out a more rich flavor. Mashing or pureeing will give a new texture. And sauteing or grilling depending on the time of year will give that yummy char!
We know that it takes some thought and effort to make food fun, but using simple tricks like spices or a new cooking preparation can really go a long way!

Sincerely,
Methia

HCU HEROES

RACE FOR RESEARCH

WALK / RUN / RIDE

HEROES IN THE MAKING

SEPTEMBER 1-30, 2023

HCU NETWORK AMERICA

aeglea

What is a virtual race?

A virtual race is a race that can be walked, ran, or biked from any location you choose. You can participate on the road, on the trail, on the treadmill (or stationary bike), at the gym or on the track (or even at another race). You get to run your own race, at your own pace, and time it yourself. You do not have to complete the miles all at once, in one day, or even a week. You can use the entire month to complete the race.

How do you know how many miles I completed?

- Our preferred method to track your miles is Strava. When you sign sign up it will prompt you to connect.
- If you choose not to connect with Strava, you can manually enter your miles.
- Please log all your miles by 11:59 pm ET September, 30, 2023

How do my miles translate to money raised?

After a racer is registered, they are set up with their own personal donation page. You can direct those who would like to donate to your race link.

Per Individual: $30

Per Family (up to 4 – 1 mailing address): $75

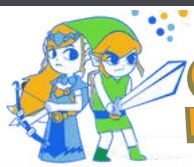
Learn more or register at https://charity.pledgeit.org/HCURaceforResearch

HCU HEROES

RACE FOR RESEARCH

WALK / RUN / RIDE

HEROES IN THE MAKING



SEPTEMBER 1-30, 2023

HCU NETWORK AMERICA

aeglea

8 Tips for a Successful Virtual Race (Tips 1–4)

#1 – Set a Goal

#2 – Recruit your Squad

#3 – Create your Training Plan

#4 – Plan the Perfect Route

Learn more & register at https://charity.pledgeit.org/HCURaceforResearch

iGive.com

You're Helping:
HCU Network America

Book Travel and earn a Donation for
HCU Network America.

Expedia

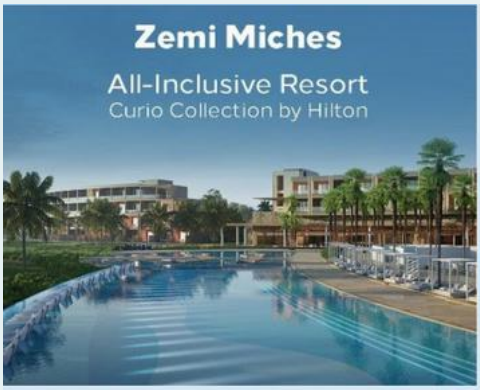
0.8% Donation

Book a rustic cabin in the woods or a luxury cabin for your summer vacay!

Hawaiian Airlines

0.6% Donation

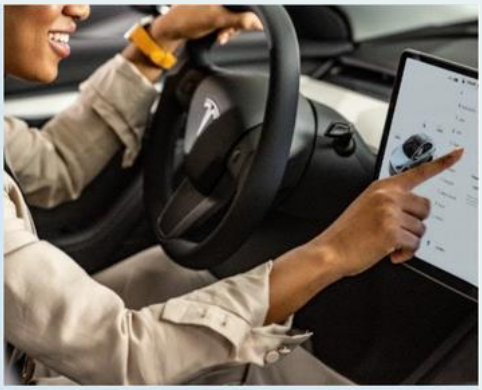
Fly Hawaiian's widebody airplanes and enjoy non-stop service options!



Hilton Hotels & Resorts

0.4% Donation

Book Hilton for All-Inclusive Escapes.



Hertz

1.2% Donation

Go electric and save up to 30%!

Traveling this summer?

Sign up with iGive to and set your charity to HCU Network America!

www.igive.com

We have travel resources!

Traveling with Cobalamin C

AN INTERVIEW WITH ADAM SETTLE

ABOUT ADAM

Adam is a 21-year-old from the Harrisburg suburbs of Pennsylvania. Adam comes from a big family, and despite being the youngest, he is the only one to have been diagnosed with Cobalamin C deficiency. Adam was fortunate to be one of the first children diagnosed through the state's newborn screening program. Despite his being legally blind, and having Cobalamin C, those who know Adam would describe him as outgoing, compassionate, adventurous, and determined.

In the book about Adam's life - No Day Wasted, the Adam Settle Story his mom summed him up in a way I think fits Adam perfectly.

"With his desire to be normal, to overcome, to not be hampered by his medical condition, Adam wanted to try everything. He got hurt along the way, but that did not slow him down. No matter how many bumps or bruises Adam endured, he quickly bounced back, ready for the next new experience…. God made it very clear to me Adam was not to be boxed up or bubbled up. He is here for a purpose, and we need to let him go."

This drive and desire to be normal and to overcome is shown in all aspects of his life, especially in his desire to travel. In Adam's book, you read a lot about the adventures he has been on with his family - Canada, Cambodia, Africa, Italy, Florida, Dubai - he is quite the traveler! Recently though Adam took his first solo trip to Indiana, so we sat down with him to catch up.

Prior to your trip did you have any worries about traveling alone?

Yes! I've always had someone with me and this time there was not my family to depend on. I was

Since you are traveling alone now, how does that change how you plan and prepare for your trip?

My parents helped me make sure I have all my medication ready to go. My dad prefilled the syringes of my B12 for the trip, typically the syringes are not prefilled and the B12, syringes and plunger are all packed separately, but in the same bag. Once all my meds are packed, my backpack that I carry on with me. This ma if they need to look at my medications.

Your bags are packed, you're go and now you are at the air Talk to me about navigating t airport - what tools are helpfu what assistance from people need.

I use an app called Aira. Unfortunately, the I airport is not partnered with them, and I didn paid version, which would have let me have f this airport.

I almost got turned around in the airport. Aft Security, I asked them where to go and they left and take the stairs. I had my backpack o my left hand, and my luggage in my right and down the stairs - but the stairs had two sets platform in between). When I got to the platf

Traveling with HCU

A REVIEW OF TRAVEL TIPS | TRICKS | EXPERIENCES

ABOUT ME

Danielle B. | Winter Park FL

My name is Danielle and I have Classical HCU B-6 Non-Responsive. I was diagnosed through Newborn Screening at 10 days old. My treatment plan consists of HCU Lophlex LQ formula, Cystadane (Betaine), Folic Acid, B-Complex and Aspirin. I live in a suburb of Orlando, FL with my husband, Irving Baez, 3 children (Irving, Julian, Christian), and 3 mini schnauzers (Mango, Bruce, Chichi). Lastly, we **LOVE** to travel!

Nassau, Bahamas (2018)

AIR

Tip: Never Check Medicine!

Many airlines allow a medical bag as an additional carry on, free of charge, as long as the bag does not contain any other non-medical items. While this is a great offer, I actually have never tried it! For air travel, I use a backpack and roller-board. If questioned, I pack my purse in my backpack which satisfies the 1 overhead/1 under-seat limitations. Formula is packed in plastic bags and when time for TSA I pull out the plastic bags filled with the formula and place in the TSA "bin". Normally, 4oz liquids do not get pulled. Due to the amount I carry, that rule usually does not apply with HCU travels.

LAND

Tip: Insulated sandwich bags

Having traveled by Amtrak, North-South Road trips, Ubers from the city to the jungle, insulated bags/sandwich bags have been the biggest formula help. They keep food fresh and pre-mixed formula cold! Pairing with my hydro flask I have no worries maintaining formula. I also bring along snacks such as dried/fresh fruit, Original Pringles (to help with altitude changes), and granola bars. Keeping it lo-pro on the road ensures room for a good dinner to celebrate at your final destination.

SEA

Tip: Bring the formula and fun!

Cruising is by FAR the easiest experience. Use extreme detail when making your reservation. I have found referencing PKU has helped a lot. I have never brought

TRAVEL PREP TIPS

Travel Hacks

- When traveling internationally, now I bring enough medication to cover an additional 5 days in case I quarantine and need to have more shipped to me.
- Have a comfortable backpack that can fit underneath airplane seat. It usually is your backpack that goes you everywhere
- Check your levels before you travel. I like doing this know where I stand in case any emergencies arise.

TOP TRAVEL EATS

Explore through food!

- 1/4 cup rice + mixed veggies is my "safe meal" for countries.
- Modify/mix-match menu items
- Eat local, it saves and usually provides fresh adapt options
- Caribbean Favs: Local fruit, Plantains, Zucchini Boa with salsa/sauces
- Central American Favs: salads with salsa, vegetari Casados, yuca fries
- African Favs: Fufu, curries, okra dishes
- European Favs: Eggless Crepes, Saffron rice

FUN MEMORIES

Click here to check out our travel & special edition topics!

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

The Homocystinurias affect multiple systems of the body!

The most common areas of the body effected are:

- The Central Nervous System (brain) 
- Ocular (eyes) 
- The Cardiovascular System (heart) 
- The Skeletal System (bones) 

But many patients experience symptoms outside of these areas!

Your quality of life matters!

Having Homocsytinuria takes a toll on our mental and phyiscal health, our our relationships, and finances. We need to hear from patients and caregivers to learn how to better support their needs.

Head to https://homocystinuria.rare-x.org/ to complete the Health & Development & Quality of Life surveys!

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

Now Available!

Betaine Anhydrous for Oral Solution 180 gm

An AB rated Generic version of Cystadane®
(betaine anhydrous for oral solution) with full
patient support services you might expect from a Brand†

Copay support

Eton Cares can help eligible, commercially insured patients get their medication for **as little as $0 per month***

Financial support

Patients who do not have insurance and meet certain financial requirements may be eligible for additional financial support from our **Patient Assistance Program***

*Restrictions, limitations, and/or eligibility requirements may apply. For patients who are not elibile for copay support or who need additional financial assistance, Eton Cares can help connect you with alternative forms of medication coverage or provide referrals to other possible sources of funding.

Eton Cares can provide copay and financial support.

Have your doctor complete the referral form to prescribe and enroll.

Click Here

IMPORTANT SAFETY INFORMATION

Warnings and Precautions

Hypermethioninemia in Patients with CBS Deficiency: Betaine Anhydrous may worsen high methionine blood levels and accumulation of excess fluid in the brain has been reported. If you have been told you have CBS deficiency, your doctor will be monitoring your methionine blood levels to see if changes in your diet and dosage are necessary.

Adverse Reactions

Most common side effects were nausea and gastrointestinal distress, based on a survey of doctors.
To report a suspected adverse event related to Betaine Anhydrous, contact Eton Pharmaceuticals, Inc. at 1-855-224-0233 or the U.S. Food and Drug Administration (FDA) at **http://www.fda.gov/MedWatch** or call 1-800-FDA-1088.

INDICATIONS AND USAGE

Betaine anhydrous for oral solution is indicated in children and adults for the treatment of homocystinuria to decrease high homocysteine blood levels. Homocystinuria is a rare genetic disorder in which there is an abnormal accumulation of the amino acid homocysteine in the blood and urine. The following are considered to be homocystinuria disorders:

- Cystathionine beta-synthase (CBS) deficiency
- 5,10-methylenetetrahydrofolate reductase (MTHFR) deficiency
- Cobalamin cofactor metabolism (cbl) defect

eTon PHARMACEUTICALS

Please see enclosed Full Prescribing Information for more information.

†Cystadane is a registered trademark of Recordati Orphan Drugs SAS, not affiliated with Eton Pharmaceuticals.

1402-v2

Acappella

NOW ENROLLING

ACAPPELLA Study on Classical Homocystinuria

Travere Therapeutics is enrolling children and adults with classical homocystinuria (HCU) in a ACAPPELLA Study. The goal is to learn more about classical HCU and the 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 participants.

Approximately 150 participants will take part 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 ACAPPELLA Study if you:

- **Have been diagnosed with HCU**
- **Are 1–65 years of age**

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

For additional information about the ACAPPELLA 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 ACAPPELLA Study. Sites are open and currently enrolling participants.

For new participants, we now have an option for the study to come to you! (decentralized site). Please inquire to learn more.*

If you have any questions, please email:

medinfo@travere.com

For more information, please scan the QR code or visit www.hcuconnection.com

*Restrictions apply

MA-PE-22-0004. March 2023

TRAVERE™ THERAPEUTICS

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://www.surveymonkey.com/r/HCUContact

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