The HCU Herald

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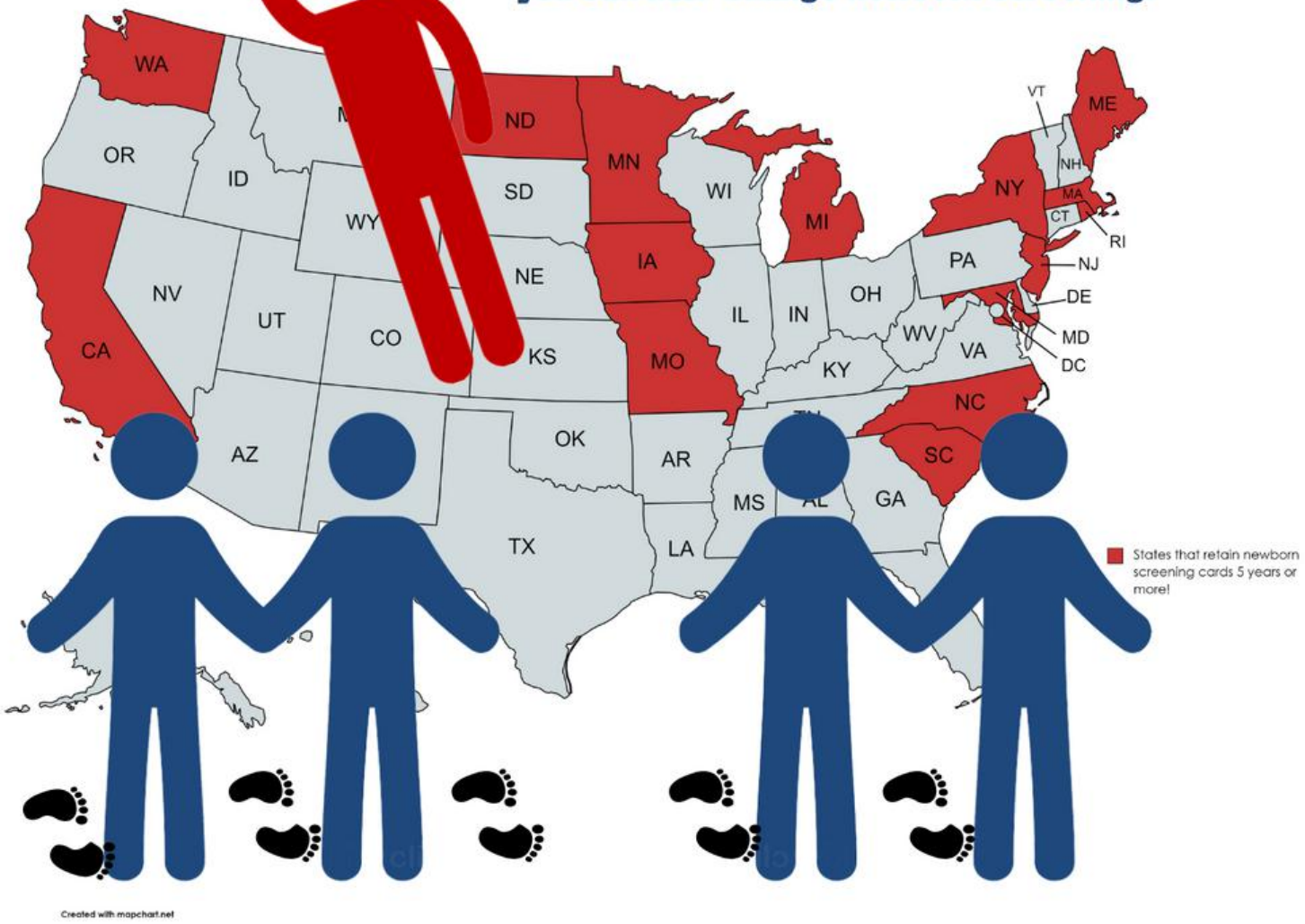
Were you or your child born in one of the highlighted states below?

Were you or your child diagnosed with Classical HCU,Cobalamin with HCU or Severe MTHFR?

If you answered yes, you can HELP change newborn screening!



Emory University is conducting a newborn screening research study on these disorders!

Contact Angela Wittenauer MSN, FNP-C, RN: alwitte@emory.edu | 404-778-8489

Director, Newborn Screening Follow Up Program | Emory Univ. Dept. of Human Genetics

HCU HERO: ANDREW FROM OREGON

In June 2020 – a bit more than 19 years after my diagnosis – it finally struck me to google "network for people with homocystinuria." Within a day, I was in the Facebook group; within a week, I joined one of the Sunday calls. And when it was my turn to introduce myself, I cried. I realized I had never met anyone else with homocystinuria, and the profound feeling of sudden connection with strangers moved me quite deeply.

In 1987, when I was 11, my mother noticed that I was crouching up very close to the posters at the science fair, and so she took me to her ophthalmologist. Sure enough, like her, I needed glasses. Over the next four years, my vision deteriorated rapidly, and soon enough my glasses were like coke bottles. At summer camp, friends would put on my glasses to pretend like they were drunk (none of us had been drunk yet – but it was how we imagined it). In 1991, when I was nearly 15, I had another check up with my ophthalmologist, and this time he felt that there must be something wrong. My vision had again gotten much worse, very fast, so he recommended we see a doctor at Walter Reed Medical Center. This doctor looked in my eyes, saw the ectopia lentis, and said to my parents, "would you like to see how far his lenses have slipped? You could probably see it from across the room." He then looked at my mother – who is 5'2" – and my father – who is 5'9" – and me – I am almost 6'4" – and said, "OK, it's either Marfan's or Homocystinuria. Let's get you over to NIH to see which."

At NIH, I saw Dr. Sandra Levin. Over the course of the spring of 1991, we made many, many trips to NIH. Typically, when I had my eyes examined, there would be a line of doctors waiting down the hallway to have their chance to look in my eyes. Once there was a team of doctors from China who took their turn. Another time they asked me if I minded answering some questions from more doctors. I said, "sure," in a classically 14-year-old way. They led me through a door to where I found myself on a stage looking out into a room of maybe 20-30 doctors, who then asked us lots of questions. (I think my mom or the doctor with us probably answered most). During this time, I also remember having a full body MRI and thinking it was kind of cool and space-age. The hardest thing about it all for me – aside from the blinding light when they took photographs of my eyes – was that I had to stop playing soccer, given the fear of contact dislocating my lenses.

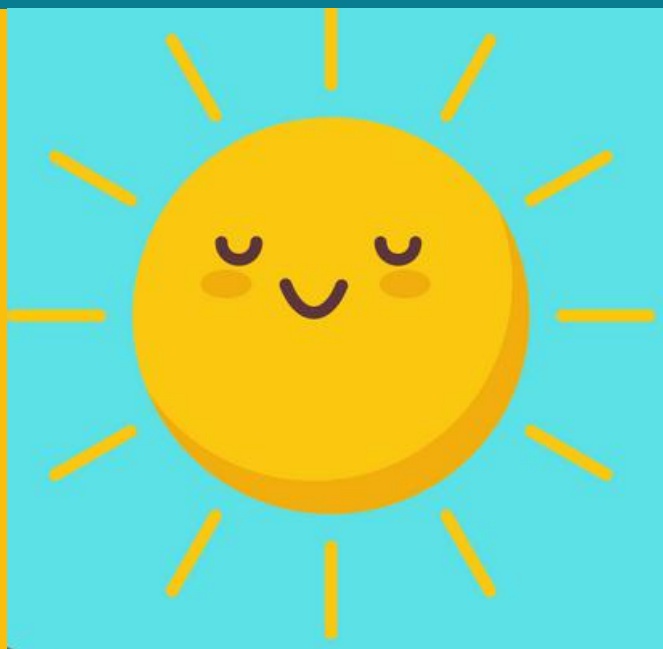
I was diagnosed with HCU. Being B-6 responsive, and generally having a mild case, I believe has afforded me a relatively healthy life. I played Division 3 college tennis – the height helps. I have earned two M.A.s and a Ph.D. I have climbed 19,000 ft mountains in Nepal, lived internationally in China, India, Bosnia and Herzegovina, and Colombia, and traveled to many, many more countries. At one point, maybe in my 20s, my mother asked me if I would trade my height for having homocystinuria, and at the time I thought, "wow…tough one actually."

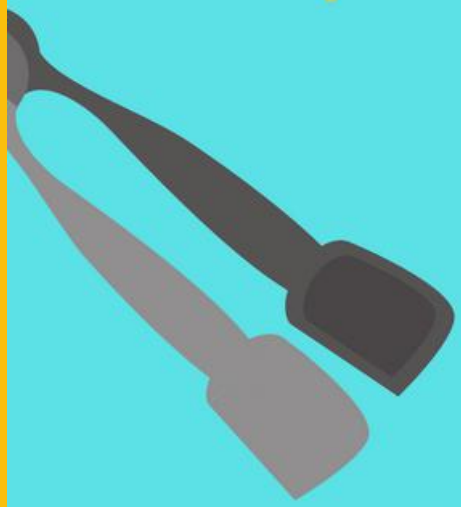
At the same time, now in my mid-40s, sometimes I feel like a Great Dane, like my body is stretched out, longer than it should be. I have had surgery on my left ankle, right knee (two surgeries), and right shoulder, and now am dealing with severe pain as a result of lumbar scoliosis. I sometimes think my body shouldn't feel like this at 45.

But this is also the body I was given, with all its "imperfections," and I have no other option but to live into it, to maximize its potential for health, to live for my 17-month-old son. Sometime in my mid-30s I think I finally matured into recognizing that I needed to truly take care of my homocystinuria. I had, before then, sometimes let the medication regime slip here and there, but from then I have been quite consistent, day and night, with the 8 little pills I take that rebalance my body chemistry to maximize my quality of life.

I am so grateful to those who participated in the early studies, to the medical professionals who investigated homocystinuria, and now to the HCU network for creating this community.

HCU COMMUNITY COOKBOOK





Hello, Summer!

Recipes at
http://bit.ly/HCUCookbook

Amber's "Steak"

Serving per recipe	3
Serving size	56 g
Protein per serving	1.6 g
Calories per serving	79

Hawaiian Jackfruit

Serving per recipe	5.4
Serving size	1/4 c.
Protein per serving	0.6 g
Calories per serving	64

Grilled Cauliflower Wings

Serving per recipe	4
Serving size	2 oz.
Protein per serving	1.7 g
Calories per serving	151

You're RARE We CARE

Introducing NEW Homactin AA Plus Powder In Refreshing Lemon Lime

Continuing to innovate for our #SmallButMighty communities!

15 PE HOMACTIN™ AA METHIONINE FREE

Lemon Lime Plus POWDER

- **Great Fresh Flavor:** Lemon Lime
- **Flexible For All Ages:** 15g PE & 150 kcals
- **Low Volume:** Mix With 5 oz Water
- **Optimized Bone Health Profile**

Barbecue Bake Medley Meal

Brookelyn Dogs

Camburgers

Get ready for summer and enjoy BBQ time with family and friends

cambrooke.com 866 456 9776

*Often we share information from our Network Affiliates - this information is not an endorsement for the product. Consult your medical team to make sure it's right for you.

HCU Network America

Eat Well, Live Well. Aji AJINOMOTO

CAMBROOKE™

NEW RESOURCES

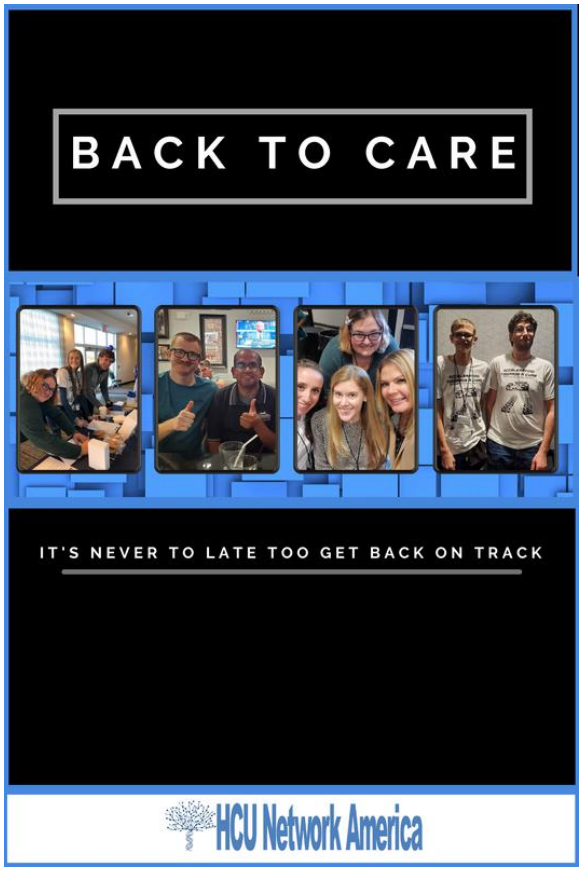
¿Hablas español?

Did you know that HCU Network America has translated 5 toolkits and guides to date this year so far? We strongly believe in health equity and one way of trying to achieve this goal is to remove barriers to education on how to manage HCU and related disorders.

To date we have translated our:

- HCU Network America brochure
- Caregiver Guide to HCU (classical HCU)
- What should a clinic visit look like? (classical HCU)
- New Family Diagnosis Guide for Cobalamin Disorders
- Emergency preparedness toolkit (all homocystinurias – classical HCU, Cobalamin, MTHFR)
- Parent Handbook for Special Education Services

In addition to this, we have linked many of our network affiliate resources to our website, and if they have them in Spanish, we have included them as well! Check out https://hcunetworkamerica.org/toolkits-and-checklist/



Back to Care Guide

Last month we announced the launch of our Back to Care Website. This month we are excited to announce the accompaniment Back to Care guide! This tool is designed to help our classical homocystinuria patients needing a low protein diet navigate their way back to diet It may also be useful for HCU patients who have been continuing with diet or newly diagnosed. The guide provides insight and guidance on how to reestablish care, provides a reminder of what HCU is, gives a diet (including formula) 101, and shares ideas on how to create a support network to help guide them through this new chapter in their lives! The guide is closed out by a letter from a patient below.

https://hcunetworkamerica.org/wp-content/uploads/2021/05/Back-to-Care-Guide-Final.pdf

SPECIAL EDITION TOPIC: BACK TO CARE, A PATIENT EXPERIENCE

Back to Care - a Patient Experience, by Danae' Bartke

It's not that I ever made the intentional decision to go "off diet", it's just that I never really was on diet to begin with. There were times where I was closer to being on diet though, than others, but it still was a far cry from how the diet really should be done. Why you may ask was this the case, well let's start from the beginning.

Diagnosis/ Middle School

In 1995, at the age of 10, I was diagnosed with homocystinuria, shortly after my younger brother was diagnosed. We were trialed on B6, then the addition of betaine, but it was finally decided we needed to follow the low protein diet and formula. We were told to aim for 15 grams of protein from food and to take our formula, which at the time was Hominex-2. We weren't really given much advice or education beyond that. We struggled for a very long time with the diet – in middle school my diet was pretty much sugary cereal, Cheeto's, Budding products (a company that sells very thinly sliced meats so they are 1 g per slice) and pop (Midwest term for soda) – I could fill up and not go over my protein. It was easy to turn to prepackaged foods because the protein content was listed – vegetables and fruit didn't come with labels, so they were this mystery of unknown protein content. Formula on the other hand was a much larger battle – it tasted awful no matter what we did to it. We mixed it with pudding, apple sauce, Jell-O, smoothies – you name it, we tried it, and I was not having it. In middle school though my aunts cornered me and told me that if I didn't take my formula they were going to either force my mom to get me a feeding tube or call Child Protective Services. The thought of a feeding tube or being removed from my family scared me into compliance with my formula.

College/Working

The years went by and having finished my associate degree, it was time to transfer to a 4-year university. As college progressed, my class load became heavier, and I worked more to offset the cost of the classes. I would work 3-12-hour days during the week, cram my classes and assignments into the other 2 days, then work another 16 hours on the weekends. With little spare time, I found myself eating out quite often. When I was with friends between classes, if they offered me food, I would take a bite of a sandwich (with meat) or eat a slice of pizza with cheese. I was so busy that I wasn't thinking of the consequences. By the time I graduated college I was eating regular cheese, bread, pasta and because Hominex doesn't travel well, I was taking it less regularly. Most days I was lucky to finish half. My last semester of college I ended up having emergency gallbladder surgery. I no longer could take my Hominex because of the large caloric load. Our clinic was able to get HCU Express 15 covered – it really turned out to be a blessing because it was a lot more convenient; it was a much smaller volume and came pre-mixed in individual servings. With the switch, I was able to get in two of the three required servings per day.

In December 2007 I graduated college, but my crazy schedule did not stop. I was motivated to find a teaching job, so I applied in multiple districts as a substitute teacher. Subbing didn't pay very well, so I also had a job in the evenings and on weekends. Similar to my time in college, I was working so much

it left little free time. When the new school year started up and I hadn't found a teaching job, I decided to find something that was more consistent and accepted a job teaching preschool in a daycare facility. The hours were consistent, the pay was better, but it would still require me to drive a long way and I'd still need to work on the weekend. Not too long after starting the hours changed. I found myself having to leave home at 5:30 a.m. to get there by 7 a.m. My workday would not end until 6:15 p.m. It made for very long days and I knew something had to change. After a year of working at that location, I accepted a job transfer to a center closer to my home. It was a small pay cut, but I also got back 2.5 hours a day of free time that I had previously spent driving. At this point, I had fallen so deep into my bad food habits, it never even crossed my mind to use that time to reevaluate what I was eating and cook healthier meals.

Blood clot

In September 2009, I started having pain in my hand and arm. I went to a clinic and the first doctor told me it was a hang nail. I knew it wasn't a hang nail, but wasn't sure what it was, so I just dismissed it and went on with life. A week later my hand turned blue, then white and very cold – I knew that it wasn't normal so back to the doctor I went. This doctor agreed it wasn't a hang nail, but wasn't sure what was wrong, so he referred me to a hand specialist. The hand specialist immediately recognized it was a blood clot and referred me to pulmonology. I called to make the appointment with pulmonology and they said they couldn't see me for two weeks. It was at that point I finally called my genetic counselor. I told her what was going on and she was flabbergasted and told me to find a ride and come to the hospital that evening. She told me they would have a specialist there to meet me that had expertise in homocystinuria and could address this.

I went to the hospital and met the specialist in the ER. I spent a week in the hospital. I wasn't allowed to do much of anything. If I wanted to eat, I had to have someone feed me. If I had an itch, someone had to scratch my itch for me – they were so afraid any movement would dislodge the clot and cause it to be fatal. After surgery to remove and break up the clot, I had to spend a couple more days there to ensure the left-over pieces didn't cause issues. While in the hospital bed recovering, I realized how lucky I was and that I needed to change my lifestyle.

Back to Care

After the blood clot, I had to be monitored closely by Hematology. I felt like I was always at the lab. They did little though, other than prescribe blood thinners, to address the real heart of the issue – my diet.

Two weeks after being released, I received an invitation to a cooking class hosted by the PKU Organization of Illinois. In one cooking class I learned more about the low protein diet, than I had acquired in the 14 years I had been diagnosed! It felt like there finally was some real education and advice that I had lacked for so long. I quickly started to change my diet. Between the hospital and cooking class, I had already stopped taking bites of things like a burger or sandwich and stopped eating regular cheese. After the cooking class, I quickly made the switch from regular breads and pasta to the low protein versions.

I continued going to events put on by the PKU Organization of Illinois, and at one of their meetings I met Malathy, owner of Taste Connections. I even traveled to Michigan and Iowa to attend her cooking classes. I finally felt like I had options that were not just low protein, but also tasty! I later found Cook for Love and that opened my options even further! I don't think I've ever made a recipe from Brenda

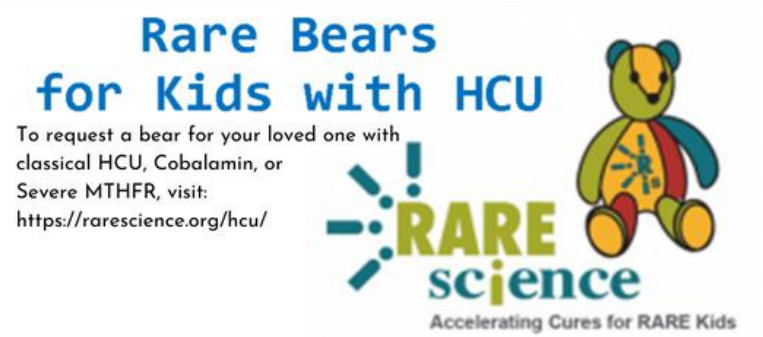
Winarski, founder of Cook for Love, that wasn't anything but delicious! I loved cooking before, but now I could cook for myself knowing these recipes were not only delicious, but low in protein.

Throughout the diet transformation, I still struggled with my formula consumption. It just never tasted good and was always clumpy. Then in 2011, I met my now husband. His passion for fitness rubbed off on me, and I started working out, but struggled with weightlifting and extreme muscle fatigue. It would take over a week to recover from a work out -even after working out regularly for a few months. He had always been great about helping me with the food part of HCU but hadn't really taken much of an interest in the formula portion. It wasn't until he put two and two together that he realized my body wasn't getting enough energy to recover because I wasn't drinking enough formula. He's the one who actually got me to start taking my full dose of formula. Once I started taking the full dose, muscle recovery wasn't really an issue anymore. I was able to quickly lose weight and build muscle.

It's been almost 12 years since my blood clot, and I haven't had any events since. I still struggle with motivation to take my formula but have learned subtle ques that my body needs it – such as brain fog, headaches, fatigue – something that was just a constant part of my life before. I get in food ruts, but then I realize it's time to look at vegetarian/vegan recipes and start trying to adapt them. When I first started my journey back to diet my levels were over 150. The more I cleaned up my diet and the more adherent I was with my formula and betaine the better my levels became. My total homocysteine now is typically 17-20. Being consistent with the diet provides me the best health outcomes and allows me the opportunity to be present for my husband and 2 year old daughter. I'm thankful that I had a second chance and was provided the guidance and education to make my way back to diet.

Pictured, Danae's husband Matt, their daughter Dana, and Danae'

RARE BEARS 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

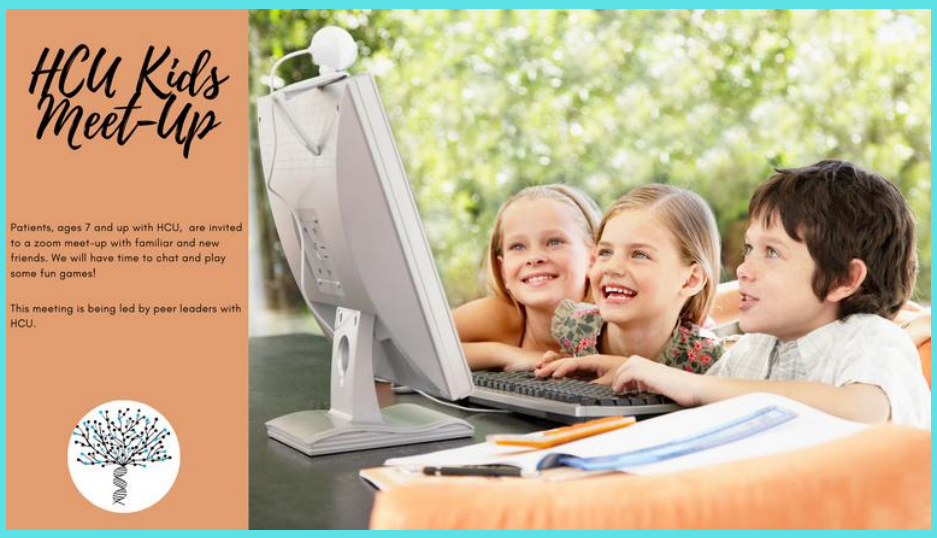
COVID IMPACT SURVEY

It's been quite a year: do you have 3 minutes to help your community? As we move toward reintroduction of the Medical Nutrition Equity Act in Congress, we are particularly interested in how COVID-19 may have affected your access to medical care and treatment for your (or your family member's) Inherited Metabolic Disorder (IMD). US Residents, please take this short survey, the results of which will be shared with the community. Thank you!

http://pheed.me/2021covidimpact

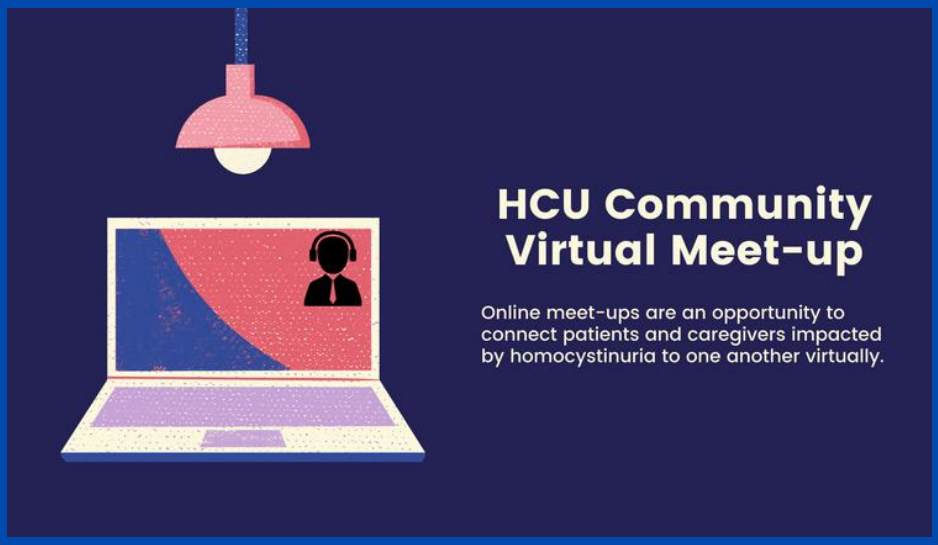
UPCOMING EVENTS

To learn more or register, visit: https://www.eventbrite.com/o/hcu-network-america-30163980100



Patients, ages 7 and up with HCU are invited to a zoom meet-up with familiar and new friends. We will have time to chat and play some games!

Come Join us!
June 15 at 2 pm CT | 3 ET
July 12 at 2 pm CT | 3 ET



- Struggling with the diet and formula?
- Feeling in a food rut?
- Don't like your formula, or having trouble getting it covered?
- Having health issues you aren't sure are HCU related, or just part of being an adult?

Come Join us!
June 6 at 2 pm CDT | 8 pm UK
June 28 at 1 pm CDT | 7 pm UK

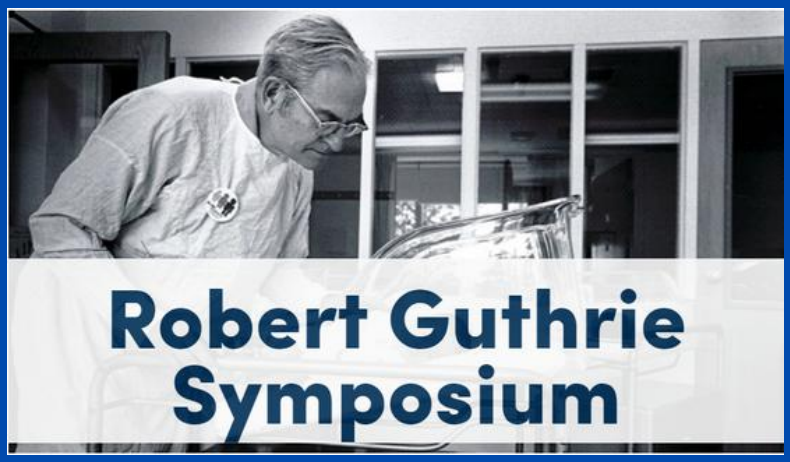
- Struggling with your b12 injections?
- Are you unsure if symptoms are related to Cbl, or just part of life?
- Curious what other therapies may be helpful?

Come join us!
June 27 at 7 pm CDT| 8 EDT

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

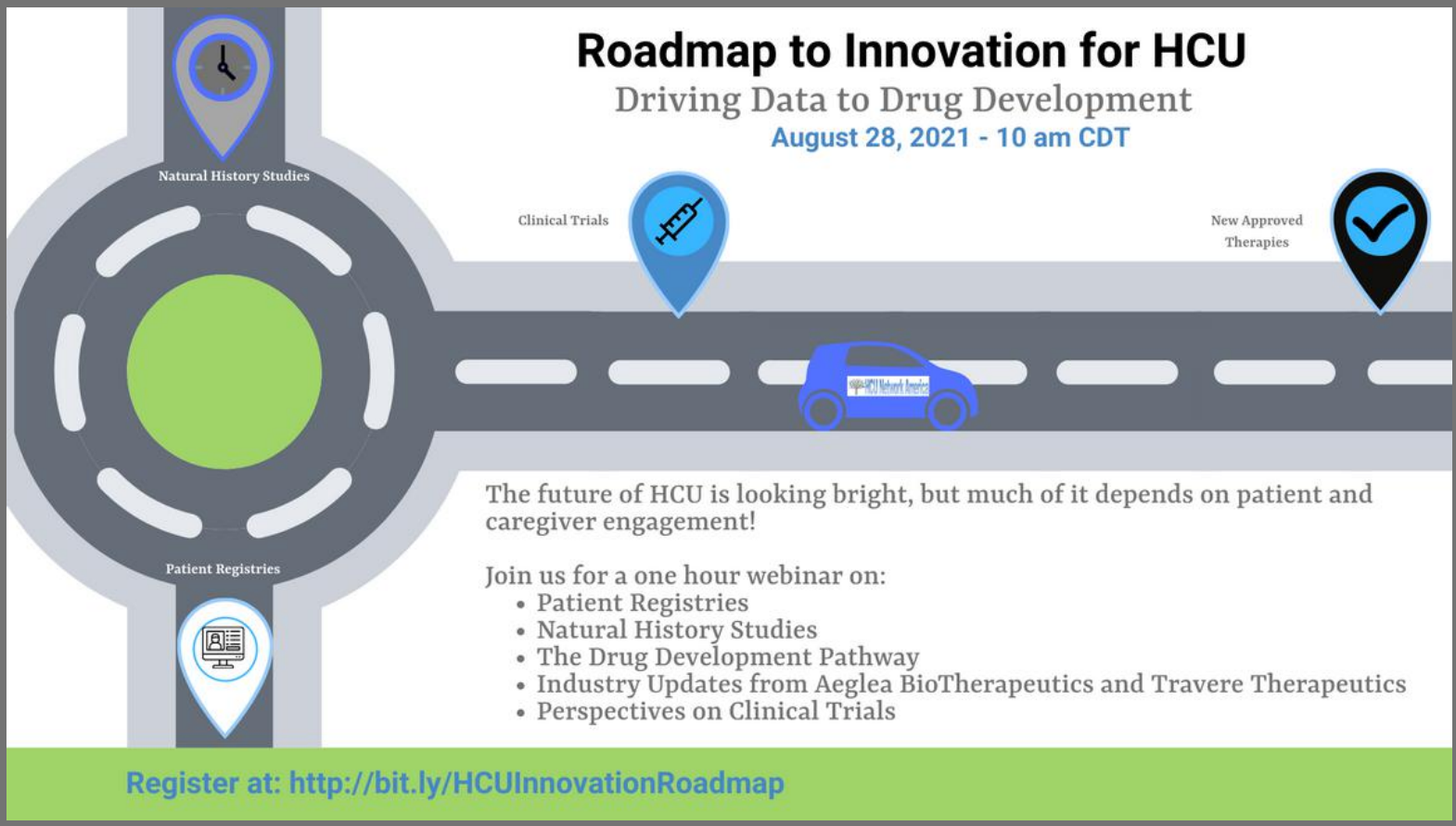
UPCOMING EVENTS

Click the image to learn more or register



The Guthrie Symposium on newborn screening and metabolic disorders is a half-day educational event honoring the life and legacy of Robert Guthrie, MD, PhD, and his attachment to the University at Buffalo and Oishei Children's Hospital and the advent of newborn screening.

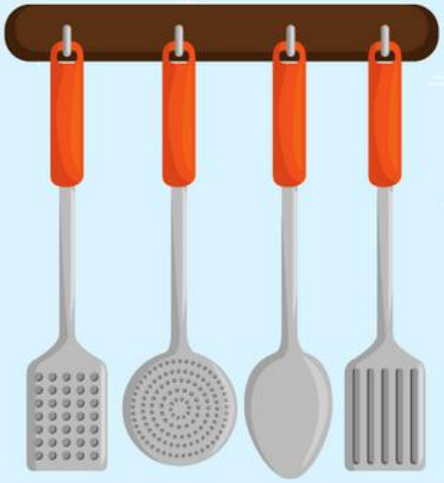
June 28, 2021 | 8 am to 12:30 pm EDT
Register here



To learn more or register, click here

LET'S GET COOKING

Cooking Demos for the Homocystinuria Community with Chef Amber Gibson



Free low protein samples to complete recipes!

*Samples only available to those in the US

We encourage children with HCU to also participate in this event!

Nutricia
Sunday, June 27
1 pm EDT

Cambrooke
Sunday, July 25
1 pm EDT

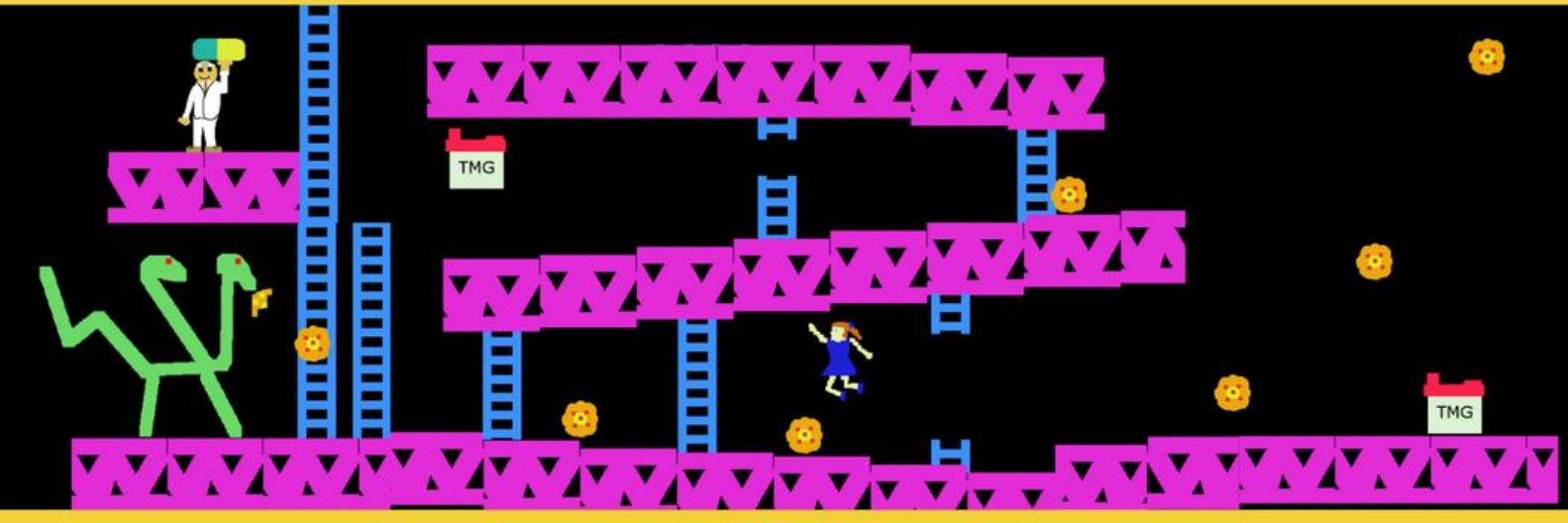
Flavis
Sunday, August 22
1 pm EDT

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

Help Henrietta rescue the cure from the Homocysteine Hydra!

RACE FOR RESEARCH

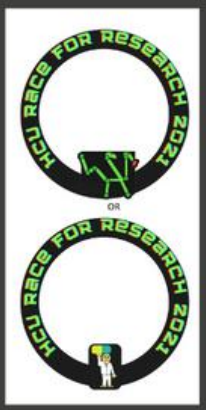
Walk, Run, Ride | September 1-30, 2021



Per individual: $30

Per Family (up to 4): $50

Registration: Opens June 1, 2021 | https://runsignup.com/Race/IL/Batavia/HCURaceforResearch



HCU Network America

RECORDATI RARE DISEASES GROUP
Focused on the Few

Focused on the Few™...

At Recordati Rare Diseases, 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.

We are proud to support the mission and vision of the HCU Network America.

RECORDATI RARE DISEASES GROUP
Focused on the Few™

www.recordatirarediseases.com/us
@RecordatiRareUS
NP-RRD-US-0231

GET INVOLVED

Rare Disease Week on Capitol Hill 2021
July 14th to July 22nd

New dates, new format... same life-changing experience!

Rare Disease Week on Capitol Hill brings together rare disease community members from across the country to be educated on federal legislative issues, meet other advocates, and share their unique stories with legislators. While we had hoped to host this event in-person, for the safety of all advocates, Rare Disease Week will be going virtual for 2021.

Registration now: https://everylifefoundation.org/rare-advocates/rare-disease-week/

Want to learn more about becoming a leader in the #newbornscreening community. Register for this free online training to develop your leadership, advocacy and storytelling skills in the newborn screening community.

Sign up for free at
https://expectinghealth.myabsorb.com/?KeyName=NavigateNBS_HCUNA#/login

#NavigateNBS

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) under grant #UK5MC321

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*

BECOME A STATE AMBASSADOR FOR HCU NETWORK AMERICA

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

HCU Network America