The HCU *Herald*

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Focused on the Few® ...

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

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
@RecordatiRare
NP-RRD-US-0201

HCU HERO: ART FROM IRELAND

A Superhero like Art

Art was 10 days old when we got the call. A man on the end of the line, a doctor from the hospital, told us there were abnormalities in our newborn screening results. The newborn screening was something you just did, never thinking your child would have one of the conditions they tested for. I asked him to spell it out, H-O-M-O-C-Y-S-T-I-N-U-R-I-A, I wrote sideways on the page. Then I started writing frantically as he started saying words like metabolic, homocysteine, methionine.....words I rattle off now. The hardest part was bringing Art in to get another blood test. He was so small and crying so hard as they stuck the needle in; my own tears fell in heavy drops onto him. I tried to comfort him but really his tight little fist was comforting me. That was the first of daily blood tests through a needle for over a week. The next day we were told the blood results still had abnormal readings and to get packed and head into Temple Street University Hospital. I felt extremely lucky we only lived a 30-minute drive from the National Inherited Centre for Metabolic Conditions at Temple Street. It was during the height of Covid, September 2020, so only one parent was allowed with Art at any one time. I was breastfeeding so I stayed with him and my husband was only able to spend an hour here and there with Art while I slept in the car. It was a challenging week. I was recovering from just giving birth, had a newborn baby, had phlebotomists coming in daily to take his blood, had a social worker, psychologist, doctors and nurses and, of course, dieticians coming in to introduce themselves to explain their role and to support me with the diagnosis of Classical Homocystinuria, a lifelong condition. I was so exhausted I could barely take in what they were saying. I remember holding Art so close that week, my tiny unique baby, as I tried to make sense of the diagnosis. Everyone was kind and empathetic and telling me how gorgeous my baby was, which was helpful as a first-time mom who just ached to go home, stare at my beautiful baby and wish none of this was happening. When Art was asleep in my arms, I would read the HCU leaflet they gave me and then copy down paragraphs from it on to a notebook to try and become articulate in explaining it. The language of the condition was so foreign to me. It was a personal heartbreak for me that I had to stop exclusively breastfeeding Art. I needed to try and control his natural protein; although I could continue to breastfeed, I needed to make sure he took his special synthetic protein formula from a bottle first. When we came home this definitely proved to be a challenge as Art would cry and cry for the breast over the bottle. We later found a dummy/soother

worked wonders to soothe him over giving him the breast. The fear was he would take too much natural protein and not any of his synthetic protein, causing a rise in the levels of the harmful substance homocysteine.

Around 1 in 65,000 babies born in the Republic of Ireland has HCU. It is an inherited genetic condition meaning Art's father and I are both carriers of the gene and the meeting of two carriers is very rare. Added to that, there was a one in four chance we would pass the HCU gene to our child. Essentially, for those of you not familiar with the condition, Art is missing an enzyme called CBS. This means he is unable to break down the amino acid homocysteine, which we all make from methionine in our diet, which causes raised levels of homocysteine., which can be dangerous. If not controlled, the buildup of homocysteine can impact the major organs of the body.

Art will "not grow out of it" nor can he eat meat, which does include chicken, and fish. He can have protein, but essentially everything that has protein must be measured to ensure it is within Art's allowance for daily protein. Bread has protein, cereal has protein, potato has protein, what you learn is everything has protein!!! Of course, Art can eat an abundance of certain fruit and vegetables. Spinach and sweet potato and banana are limited, but he has no interest in eating an abundance of fruit and vegetables for us! A big achievement was, after 16 months, going from weekly blood tests to fortnightly or every 2 weeks visits. We go to Temple Street to get the blood taken, and the phlebotomists are now like old friends and it makes such a difference that we get a big welcome when we see them. We then call the dieticians to get the blood results and discuss them before making amendments to Art's diet as required. For example, lowering his intake from 6g of protein to 5g when his homocysteine levels spiked. Or increasing his intake from 6g to 7g of protein per day as his natural protein levels of methionine were too low. As it is a metabolic condition, anything like teething or a bout of sickness can cause the homocysteine levels to rise. It took well over a year before I started to not have a reaction close to despair when we received results of raised levels. I began to learn that it is the overall average levels over a number of weeks and even months that mattered and once they were at a good level and Art was developing well, there were no concerns. However, it is a constant balancing act between providing enough protein for Art to grow and curbing intake and ensuring he gets his synthetic protein to manage the homocysteine and other amino acid levels in the blood.

We attended a clinic initially every 3 months and now every 4-5 months. At the clinic we meet Art's metabolic team, the dieticians, nurses, the NCHD's (non-consultant hospital doctors) and his consultant. He is weighed, his height is measured and his development checked. The team of dieticians in Temple Street are supportive. They are there when we need them to be to allay our fears and concerns and to remind us that our child is developing well and meeting/exceeding all his milestones! Our consultant, who will be Art's consultant until he turns 18, is a straight talking, matter of fact woman whom we love. She always fields my realms of questions with good humor and provides to-the-point answers, which I love. When you get a diagnosis of such a rare condition, having someone who can explain it simply and with

clear language is a god send. We found the community on HCU Ireland Facebook page extremely helpful and of course HCU Network America who actually put me in touch with HCU Ireland! You realize what a small global community HCU patients are, and it is wonderful to feel connected in that global way. The other mothers just totally understood how I felt, and I can always ask for some tips or advice as they've been through it all. Recently, Art has been going through a challenging toddler eating stage that all toddlers go through and a growth spurt which means his natural protein intake has increased. Therefore, we often find and measure the bits of waffle he left on the plate and floor trying to figure out how much he actually ate. When he mashes the food between his fingers, we give up! I posted on the page asking for tips and got great suggestions. I find the low protein recipes on Vitaflo very helpful and what_esmee_eats on Instagram. The bread recipe on Vitaflo is one we turn to weekly.

One big battle for us was obtaining a medical card for Art. Again, HCU Ireland mammies were so helpful here and guided us through the options. However, as it is such a rare condition there are big divergences in what everyone has received in terms of state support. We applied and received Domiciliary Care Allowance which is a monthly state grant that also provides an automatic medical card. We would be happy to speak to anyone thinking of applying for the DCA. The DCA is invaluable support with our monthly shopping bills. Our cost for food tends to be high as a result of purchasing food not on prescription but that we need in order to add variety to his diet and up his protein levels such as gluten free bread, oat milk and vegan cheese, items that tend to be pricier. In addition, with his medical appointments, I felt I needed to reduce my hours at work to try and manage these.

One thing I would say to new parents of a child with HCU is that you will learn more about it week by week and before long you are an expert. You will know all those terms and words, and you will one day feel a bit of that weight off your shoulders as you realize it is under control. You'll learn to read labels to check protein content in 2 seconds flat in the supermarket to see if your child can eat it and you will be able to look at a piece of avocado and say that's 50g! However, you need to be patient with yourself as it is a journey, and a lifelong one. It also helps that my husband Dave and I work as a team, and we try our best to be patient with one another knowing there will be inevitable ups and downs on this journey.

Art is now 18 months old, his favorite book is called "A superhero like you" and he certainly is our superhero! He has met all his milestones; he was walking before 12 months and has been talking since he was 16 months old. He LOVES, LOVES the guitar. We once showed him a Bruce Springsteen acoustic video and now he asks for "Bruce" all the time! Who knows, maybe he will become the first HCU global rock-star? He is full of energy, love, hugs, and fun. We feel extremely lucky with our unique, rare baby and grateful for newborn screening. We are hopeful for a future where there may be treatments other than a strictly controlled diet and blood tests for Art. We are so hopeful, in fact, that we hope one day to expand our family. This may mean we have another baby with HCU (1 in 4 chance), but if they are anything like our Art why wouldn't we take the risk?

If you wish to get in touch with HCU Ireland, check out the Facebook page: Homocystinuria (HCU) Ireland or email hcuireland@gmail.com

THIS WEEK'S MENU

Each day has meals for <10 grams (g) of protein/day, 20-30 g. of protein/day, and 30-40 g. of protein/day.

M

Breakfast: Toast & Yogurt
Lunch: Spaghetti & Broccoli
Dinner: Pizza

T

Breakfast: Cereal & Fruit
Lunch: Tostada
Dinner: Grilled Romaine & Pasta

W

Breakfast: Avocado Toast
Lunch: Vegetable Soup
Dinner: Eggplant Rolls

T

Breakfast: Pancakes
Lunch: Cauliflower Fried Rice
Dinner: BBQ Sliders & Jicama Salad

F

Breakfast: Banana Bread & Spread
Lunch: Tortilla Soup
Dinner: Zoodles & Marinara

Shopping List

Click each day to view the week long menu!

Disclaimer: This meal plan is intended to be a foundation or guide to what meals could look like on a low protein diet. It does not take into account individual caloric, protein and formula requirements, which are all patient-specific. Please consult with your metabolic geneticist and dietitian prior to making any significant dietary changes or following any meal plans of which you are unsure.

Baked Feta Pasta

Yields 3.6 servings
Serving Size: 6 oz.
Protein per serving: 0.9 g
Calories per serving: 421

Ingredients:

- 140 g Low-protein Fusilli
- 8 1/8 oz. Violife Just Like Feta Block
- 10 tomato(s) Grape tomatoes
- 2 TBSP Olive Oil
- 1/4 tsp Italian Seasoning
- 2 clove(s) Garlic, minced
- 2 tsp Balsamic Vinegar

Directions:

1. Preheat oven to 375 degrees. Cook pasta according to package directions.
2. While pasta is cooking, place feta block in the center of an 8x8 baking pan. Spread tomatoes and minced garlic around the feta. Sprinkle Italian seasoning over the feta and drizzle with olive oil. Place in the preheated oven and bake for 15 minutes. Remove pan from oven and gently mix the feta and tomatoes together to create a sauce. Add the cooked pasta and balsamic vinegar and gently toss to combine. Season with salt and pepper to taste. Serve!

Notes: Add crushed red pepper for a little extra bite.

You can add canned artichoke hearts to the dip also. Adjust the protein as necessary

MEET OUR MEDICAL ADVISORS

Abby Hall, RD

Abby Hall is a metabolic dietitian in the Medical and Molecular Genetics Department at IU Health Riley Hospital for Children in Indianapolis, IN, United Sates. Abby has been a dietitian for 12 years and has specialized in inborn errors of metabolism, including homocystinuria and other methylation defects, for the past nine years. She holds a Bachelor of Science degree from Indiana University with a major in Nutrition and Dietetics. She then completed her dietetic internship with a focus in community nutrition at University of Buffalo – part of the State University New York (SUNY) system in Buffalo, NY. After her internship in 2009, Abby moved back to her home state of Indiana and began practicing as a licensed and registered dietitian in a position focused on adult and pediatric nutrition support. During her internship and first job, Abby discovered a passion for working and caring for pediatrics and a desire to specialize in a specific area in nutrition. When the opportunity arose to join the metabolic team at Riley, Abby jumped at the opportunity and has never looked back.

Through her time as a metabolic dietitian at Riley Hospital for Children, Abby has had the opportunity to work with an extensive variety of metabolic conditions through every stage of life. From newborns and their families, to the elderly patients who some pre-date the technologies of newborn screening, Abby has recognized the uniqueness of the field she is in and the ever-evolving care of patients with inborn errors. The special bond created with families and patients is one of the most rewarding parts of her position. One day she is walking a family through mixing a special formula for the first time, and the next she is celebrating a high school graduation. Abby truly appreciates the connection and her unique position to support and work with these families to provide the best care possible.

Janette Skaar, RD, CSP, CNSC

Janette Skaar is a metabolic dietitian at CHOC- Children's Health of Orange County, in Orange, CA. Janette specializes in medical nutrition therapy for inborn errors of metabolism, including homocystinuria and other methylation defects. She holds a Bachelor' s degree from the University of Iowa with a major in Food and Nutrition and completed her dietetic internship at the University of Iowa Hospitals and Clinics. Janette began her clinical nutrition career in 1984 as a registered dietitian at University of California, Irvine Medical Center, where she was introduced to metabolic disorders and was the primary dietitian in the PKU clinic. In 2003, Janette went on to specialize in nutrition support in neonatal intensive care and pediatric intensive care at CHOC. In 2016, she had the opportunity to join the metabolic team at CHOC, returning to her passion for working with and treating metabolic patients, both as in-patient during critical illness and in the outpatient setting. In coordination with the metabolic team at CHOC, Janette focuses on tailoring nutrition plans for individuals and partnering with their parents and caregivers to help them achieve optimal metabolic control. Janette is also involved in metabolic research studies where CHOC is a participating enrollment site. Janette has enjoyed supporting HCU Network America by joining in their virtual fundraising races, most recently HCU Race for Research!

Make some noise and help get the Medical Nutrition Equity Act Passed!

- **February 28: Share a picture of you or your loved one effected by homocystinuria with post**

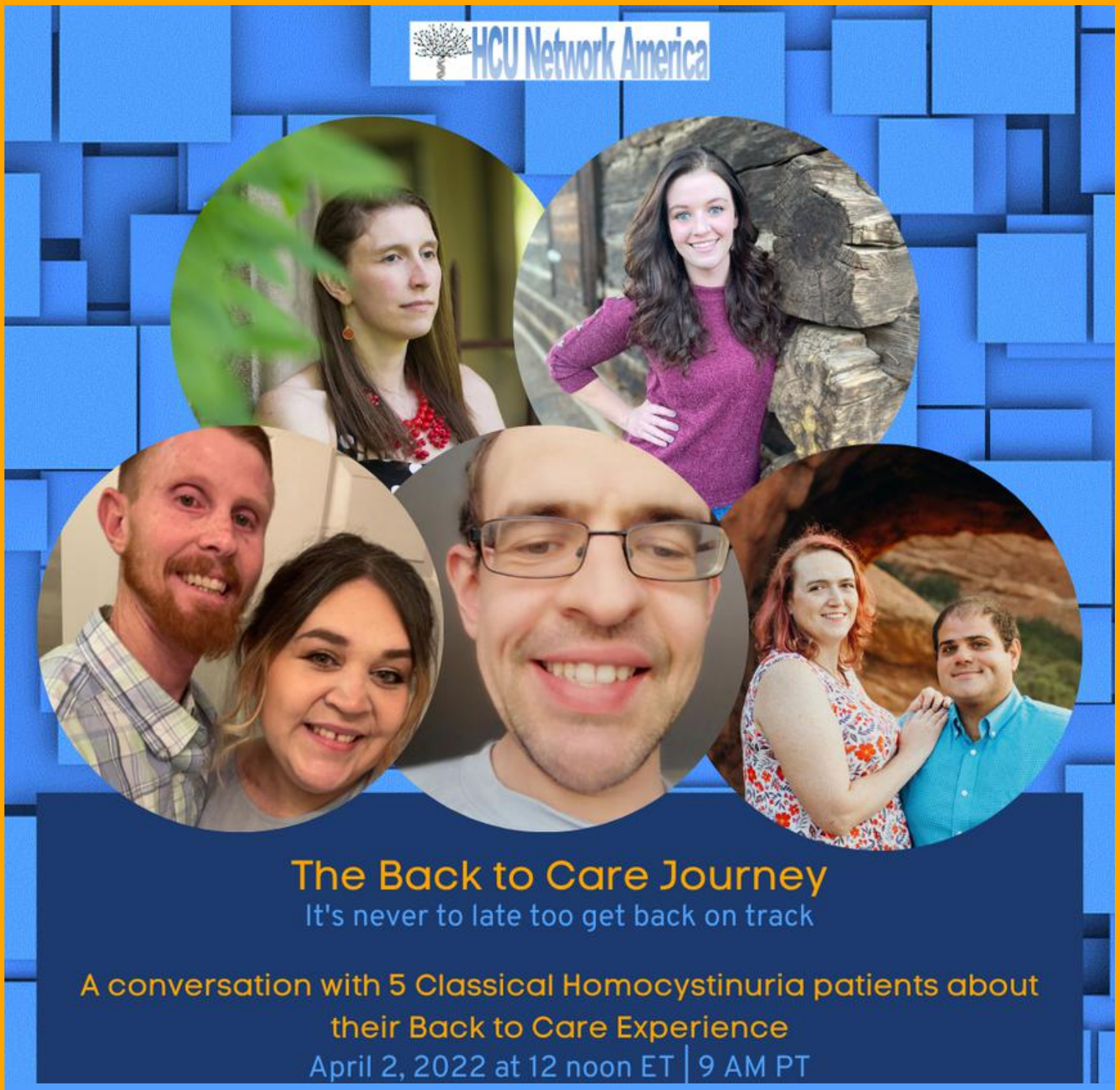
  #whynow4MNEA? Every month of my kid's life we struggle to pay for her essential #medicalfoods. Keeping her healthy is hard enough. We need Congress to pass #S2013HR3783 to ensure coverage for treatment for her disorder. #medicalnutritionequitynow nutritionequity.org

- **March 1: Share a picture of your life saving forms of medical nutrition, what it cost, and why it's important to you with the #WhyNow4MNEA and #S2013HR3783**

- **March 2: Fill out the contact congress form (it only takes a minute!)**

- **March 3: Share your patient story on nutritionequity.org**

- **March 4: Tweet at, tag on Facebook/Instagram your Congressmen, asking them to support #S2013HR3783**



We know it's easy to stray away, but as patients age, they realize the importance of keeping in contact with their clinic, following their diet, and keeping themselves all around healthy!

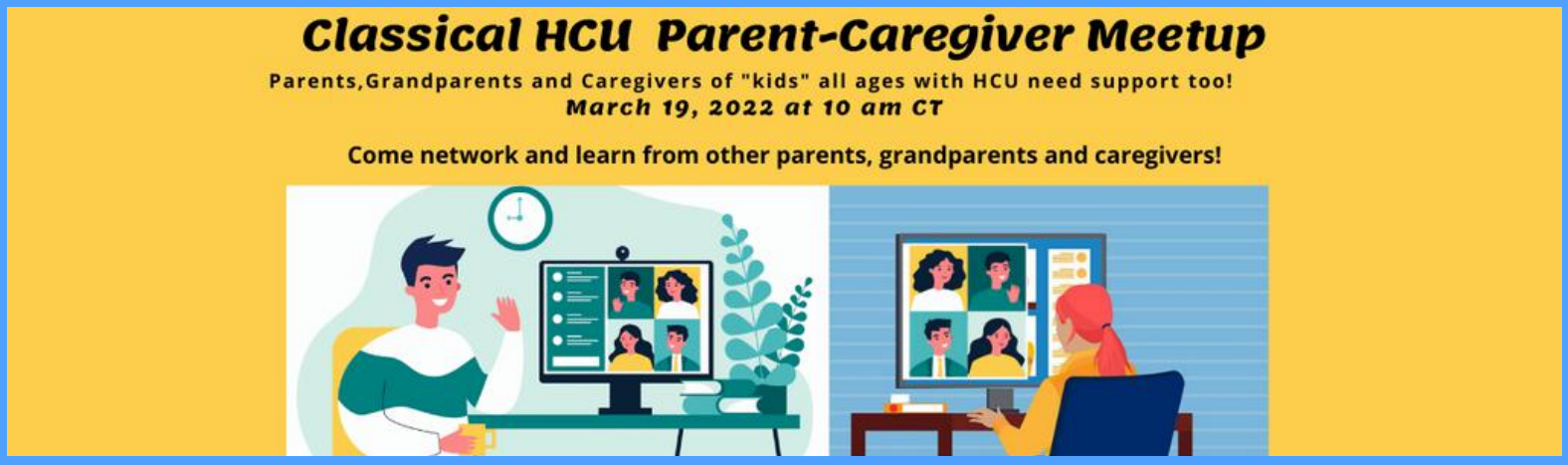
Join our patient-moderator Danae, and patients Valerie, Janet, Ashley and Aimee as they share their personal journey that led them back to care.

Register here: https://www.eventbrite.com/e/276087052807

Learn more about the HCU Network America Back to Care program at https://hcunetworkamerica.org/back-to-care

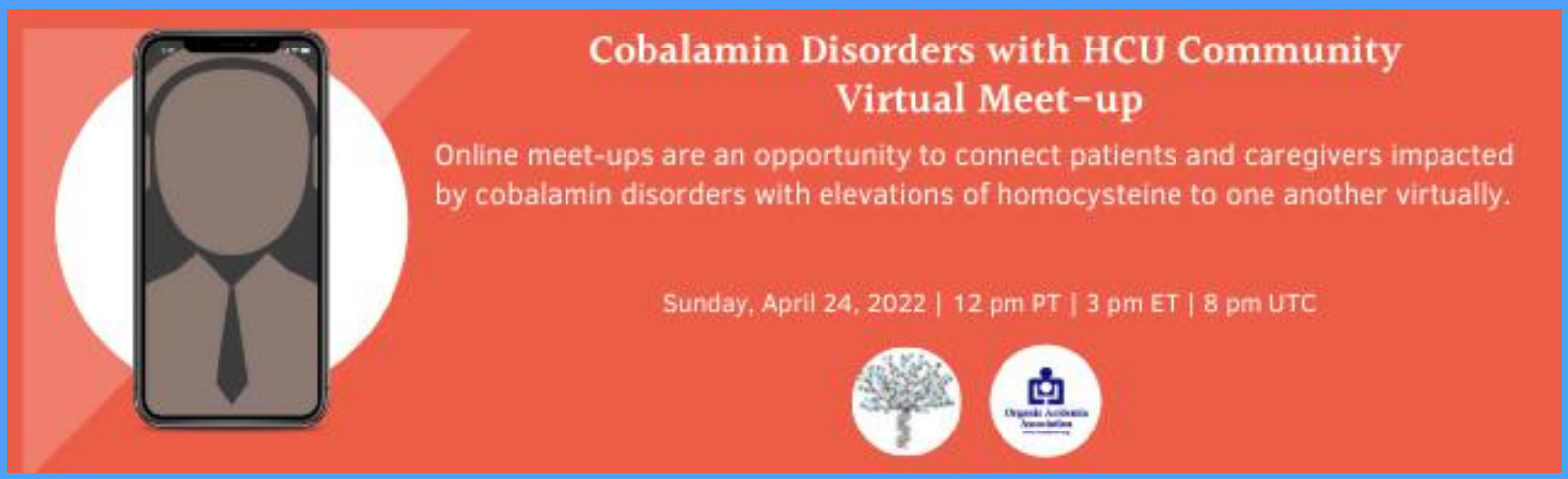
UPCOMING EVENTS

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



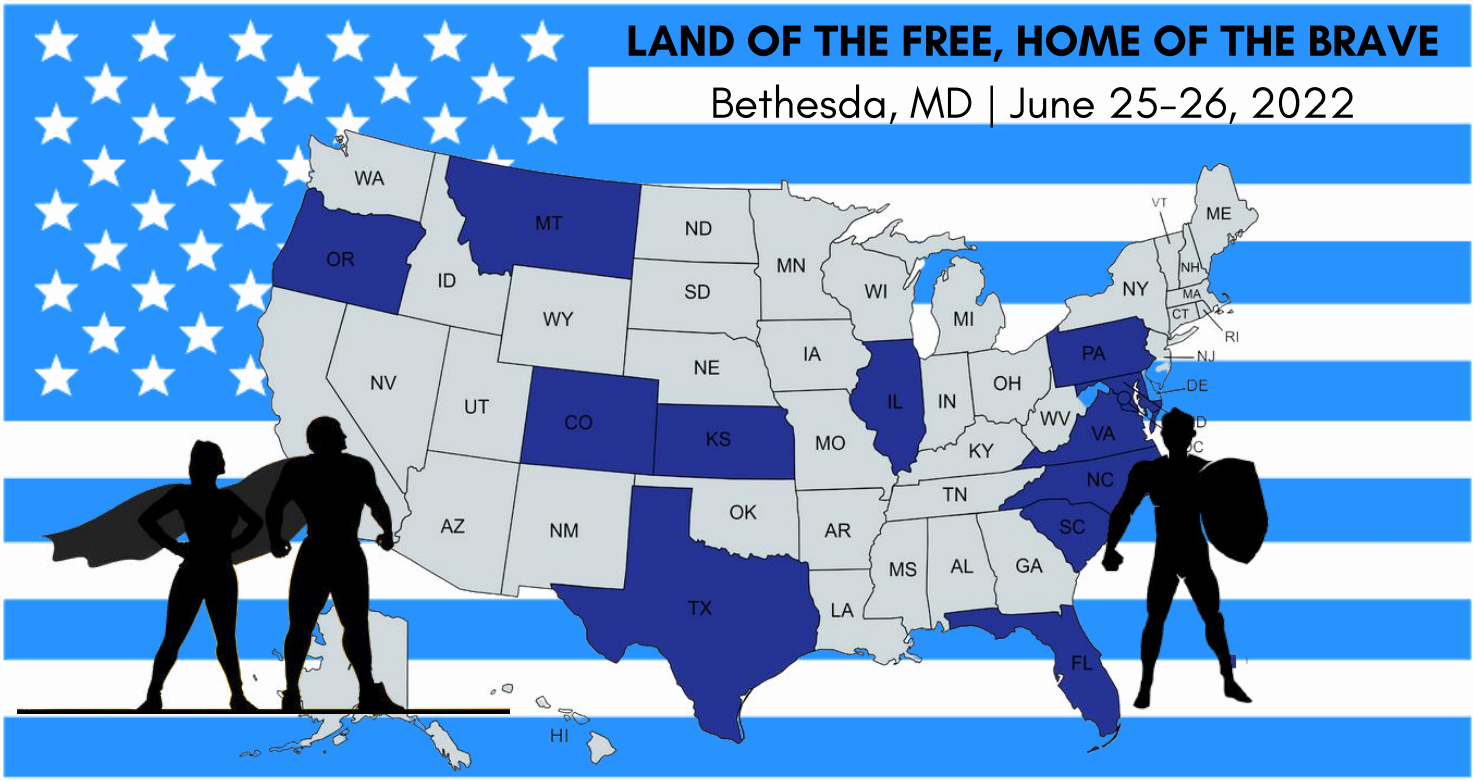
Whether your child is 5 months old or 25 years old, parents, grandparents and caregivers of those with HCU need support! Come join us for networking, tips, tricks and conversation.

Join HCU Network America and Chef Amber as she demonstrates for us 3 quick and easy low protein dishes! All low protein patients and caregivers are welcome!



Join our host Brittany Parke, a Cobalamin G mom for networking, tips, tricks and the support we all need when navigating a rare disease!

HCU Heroes Unite!



Other locations not displayed, Canada and Uruguay:

In 4 months, we will be packing our bags and heading to Bethesda to reunite with our HCU family!

Registration is open and our map is filling up – it's even gone global! Head over to our conference page and see all we have planned and to register: https://hcunetworkamerica.org/2022-conference

- *All attendees 2+ must wear a mask at all times.
- **All attendees 5+ will be required to show proof of vaccination.
- ***Attendees under the age of 5 will have to present a negative PCR dated no more than 3 days prior to the event.

Attention: Volunteers Needed

For HCU Network America, Organic Acidemia Association and Propionic Acidemia Foundations Conference:
Land of the Free, Home of the Brave
(An "educational meeting" for patients and families affected by homocystinuria, a rare genetic disease)

Pooks Hill Bethesda Marriot | 5151 Pooks Hill Rd, Bethesda, MD 20814

We are now seeking volunteers to assist in our children's program:

- Arts & Crafts
- Educational Lessons
- Stories
- Games
- Hands on activities
- And more!!!

Children's program will take place during the main session of the conference.
(Saturday 8:00-5:30 and Sunday from 8:00-12:00)

Please see available slots at: https://signup.com/go/miiZeeB
or
Direct your contact with days and times of availability to:
Danae' Bartke, Executive Director at
info@hcunetworkamerica.org

NORD® RARE ACTION NETWORK®

The RareAction Network®, powered by the National Organization for Rare Disorders (NORD), serves to connect and empower a unified network of individuals and organizations with tools, training and resources to become effective advocates for rare diseases through national and state based initiatives across the United States. We stand for equitable access to timely diagnosis, treatment and care for every person impacted by a rare disease.

Rare Disease Day may have passed, but there is still time to join the fun
NJ - 3/16 at 10am ET Register here | DE – 3/19 at 12pm ET Register here

Don't live in NJ or NE? Find your state's Rare Action Network here

RESEARCH OPPORTUNITIES

The HCU Networks (HCU Network America and HCU Network Australia) have partnered with RARE-X to drive a Data Collection Program for the homocystinurias. The RARE-X platform enables the gathering, structuring and sharing of critical patient data at scale. This patient data will help accelerate research, drive disease understanding and enable the development of new diagnostic tools and future treatments and cures.

The HCU Networks are building the Homocystinurias Data Collection Program to:

- Inform researchers how homocystinurias impact patients and change over time.
- Enable better data to use in drug development and clinical trials.
- Give patients the opportunity to participate in clinical trials.
- Reduce the time it takes to study new medicines.
- Accelerate the time to get treatments to patients.
- Enable the use of data as a placebo (instead of actual patients) in a clinical trial.

The Homocystinurias Data Collection Program is patient-owned and enabled by RARE-X technology. All data governance, consent support, and data security are delivered by RARE-X. Privacy is something RARE-X takes seriously and patient names are never revealed.

Since RARE-X is a nonprofit, there is no cost to you or the HCU community. RARE-X's mission is to serve patients and drive research towards therapeutic development.

Contribute to a brighter future for homocystinuria, register now at:
https://homocystinuria.rare-x.org

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

STRENGTH IN NUMBERS

THE FACTS

The Homocystinurias Data Collection Program (HDCP) enables the comprehensive collection of data so we can accelerate research and drive medical advancement for the homocystinuria community. Data is critical in driving medical advancements for the HCU community and your participation in the HDCP is one of the most important and critical efforts you can do. By registering, being counted, and answering questions about how homocystinuria has affected you, you can help researchers better understand the disorder and develop future treatments and cures.

WHO CAN SIGN-UP?

- Anyone with one of the homocystinurias or their caregiver.

WHAT DO I NEED?

- A computer, tablet, or phone with internet connection.
- An email address.

WHAT IF I NEED HELP?

- Access the User Guide or **support@rare-x.org** for questions.

WHAT KIND OF INFORMATION IS COLLECTED?

- Basic demographics
- Health and treatment history.
- Family history.

WILL MY INFORMATION BE PRIVATE?

- Yes. Your de-identified data will only be shared with those whom you want to have access to it, and only for reasons you determine are appropriate.

WHO OWNS THE DATA?

- Patients and caregivers who contribute data to the HDCP own and manage their data. They decide who has access to it and how it's shared.

DO I NEED TO UPDATE MY INFORMATION?

- Yes. Update annually or if symptom change. You will be notified of new surveys.

CAN I STOP BEING PART OF THE PROGRAM?

- Yes. You can stop taking part at any time for any reason.

WHY REGISTER?

ENHANCED DATA TO ADVANCE RESEARCH → **BETTER UNDERSTANDING OF DISEASE** → **ACCELERATING THE DEVELOPMENT OF NEW TREATMENTS**

→ **IMPROVED HEALTH OUTCOMES FOR PERSONS WITH HOMOCYSTINURIA**

CONTRIBUTE TO A BRIGHTER FUTURE FOR HOMOCYSTINURIA

For more information or to register visit: **https://homocystinuria.rare-x.org**

HCU Network America

HCU Network Australia
HCUNetworkAustralia.org.au

POWERED BY RARE-X

Were you missed by Newborn Screening?

WE HAVE AN OPPORTUNITY TO HELP CHANGE THE PROCESS, BUT NEED YOUR STORY TO GIVE US THE EVIDENCE TO BUILD OUR CASE

But we have newborn screening For HCU...

According to recent statistics, approximately 25-50% of patients are missed by newborn screening for Homocystinuria. There are multiple factors that can play into these numbers. Currently it is federal mandate that all states screen for Classical Homocystinuria through the newborn screening test, but there are no set standards. Meaning, every state or region can set their own methionine cut offs. A handful of states also do tier two testing—meaning they have a second round of newborn screening, making it more likely for homocystinuria to be picked up. Another factor that plays into the effectiveness of the test, is how elevated the patient's levels are at the time of the test. Patients who are pyridoxine (B6) responsive, or have more functioning CBS enzyme, are less likely to be picked up by the newborn screening

What about Cobalamin disorders and Severe MTHFR?

Cobalamin C, D, F, J and X are also part of the newborn screening process as secondary conditions. These conditions can also be missed.

Cobalamin E, G and Severe MTHFR are not part of newborn screening, they are sometimes diagnosed. What we are advocating for would improve diagnosis for all types of homocystinurias - these included!

So how can you help?

If you or your loved one were missed at screening, we need to hear from you ASAP so we have enough evidence to bring about change. Contact Danae if you can help us, and she will lead you through the process that is outlined on the next page.

Talk to your geneticist about the newborn screening survey and urge them to complete it! This will help us build support for changes to the process to increase the likelihood that HCU patients, regardless of type will be diagnosed at birth.

On the following page you will find the letter portion. We ask you to give to your clinic, followed by the survey form:

To Whom this may concern,

I would appreciate your support in answering a brief survey to help support efforts to improve newborn screening for the homocystinurias.

I have been working with HCU Network America, a patient advocacy and support group for the Homocystinurias (HCU), for whom I serve as a medical advisor. One of their key goals is to improve newborn screening for HCU, as it is estimated that over half of patients are missed by the current screening process and often are not diagnosed until they have developed serious clinical symptoms. To build support for an improved process, we are collecting information on patients missed by the current screening process, which we intend to then publish in a consolidated case report.

Could you please support our efforts by completing the attached brief questionnaire, and sending it to me vie-email at: FICICIOGLU@email.chop.edu

Sincerely,
Can Ficicioglu, M.D., Ph. D.
Director of Newborn Metabolic Screening Program, Children's Hospital of Philadelphia

Survey on Homocystinuria (HCU) Patients Missed by Newborn Screening

Do you have any patients with HCU missed by NBS and diagnosed later based on symptoms?
( ) Yes ( ) No

If yes, at what age were the patients diagnosed, and what year were they born and in what state?

Age at diagnosis (mos.) _____ Year of birth _____ State born_____
Age at diagnosis (mos.) _____ Year of birth _____ State born_____
Age at diagnosis (mos.) _____ Year of birth _____ State born_____

Would you be willing to provide information to contribute to a "Case Report" we plan to publish on patients missed by Newborn Screening?

What is the name and address of your clinic and the best contact person for further information?

- Clinic Name:
- Clinic address:
- Contact Person:
  - Name
  - E-mail
  - Phone

Please send completed survey to Dr. Can Ficicioglu at Ficicioglu@email.chop.edu
Or online: https://hcunetworkamerica.org/survey-on-homocystinuria-patients-missed-by-newborn-screening/

Compose

COMPOSE Study

A study that looks at how safe pegtibatinase is and how well it works in people with classical homocystinuria (HCU)

NOW ENROLLING

Travere Therapeutics has initiated a first-in-human study of pegtibatinase, a new, investigational human enzyme therapy that targets the underlying enzyme deficiency that causes HCU.

The goal of this study is to learn how safe and effective pegtibatinase is and how well it works in people with HCU at different dosage levels.

Approximately 32 subjects will participate in sites in the US. The study will include three key stages (screening, treatment, and extension) and will last approximately 150 weeks.

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

- **Have been diagnosed with HCU**
- **Are 12–65 years of age**

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

For additional information on criteria for eligibility, please go to: www.clinicaltrials.gov/show/NCT03406611

Payment for time and travel may be available to subjects who participate in this study.
To inquire about participation in the study, **please contact**:

HCUConnect@labcorp.com

TRAVERE™ THERAPEUTICS

MA-497. August 2021

NEWS YOU SHOULD KNOW

Generic Version of Cystadane Now Available

February 15, 2022 - The first generic version of Cystadane® (betaine anhydrous for oral solution) Powder has been made available by Oakrum Pharma, LLC, in collaboration with ANI Pharmaceuticals. The Food and Drug Administration approved the Abbreviated New Drug Application (ANDA) for Betaine Anhydrous Powder in a 180 gram bottle. The approval was granted Competitive Generic Therapy, meaning the Company will have 180 days of exclusivity to market the product.

The generic is currently available through Acaria Health. If you want to order it, have your doctor write a new prescription with the generic name and indicate that generic is preferred, and send it to Acaria Health. Call them for details. If your insurance rejects your prescription for brand name Cystadane, which they sometimes do once a generic is available, Anovo Rx will also work with you to get your script transferred over to Acaria Health.

If your prescribing medical provider has concerns about generic, your provider can oppose the change if being mandated by insurance, but it will likely result in you paying a higher co-pay.

For those who currently are getting their betaine from a compounding pharmacy, you can have your prescribing doctor fax prescriptions to 1-877-541-1503. If there are any questions, AcariaHealth's direct contact number is 1-855-422-2742.

See full press release at: https://www.empr.com/home/news/generics-news/generic-version-of-cystadane-now-available/

SCHOLARSHIP OPPORTUNITIES

Living with a rare disease means managing unique challenges, including frequent doctor visits, rigorous treatment regimens and hospitalizations, and exposure risks. While quality and duration of life continues to improve thanks to improved diagnosis and treatment approaches, individuals living with rare diseases still face disparities in achieving traditional life milestones.

That's why The EveryLife Foundation for Rare Diseases established the #RAREis Scholarship Fund – to enrich the lives of adults living with rare diseases by providing support for their educational pursuits. Thanks to the support of Horizon Therapeutics RAREis, one-time awards of $5,000 each will be granted to up to 32 recipients for the Fall 2022 semester.

Applications for Fall Semester 2022 will open March 18, 2022
Learn more and apply here: https://everylifefoundation.org/rare-scholarship/

WHAT'S YOUR STORY?

More than 30 million Americans are living with one or more rare diseases.
Each one of us has a story. We want to hear yours.

Today, the EveryLife Foundation for Rare Diseases launched *What's Your Story?*, a new online rare disease story bank for advocates like you to share your experiences on a wide-range of issues impacting the rare disease community.

What's Your Story? presents a series of questions covering topics ranging from difficulty in getting diagnosed to challenges in gaining access to therapies, to facing discrimination in the workplace, and more. You are also invited to upload photos and videos that help illustrate your rare disease journey. Advocates of any age are welcome to submit stories.

Share Your Story With Us

Benefits of sharing your story:

Influence rare disease policy.

Educate policymakers.

Inspire other advocates.

Be entered to win a $1,500 donation to the patient advocacy organization of your choice! Submit your story by March 2nd, the final day of Rare Disease Week on Capitol Hill 2022 for the chance to win one of three donations.

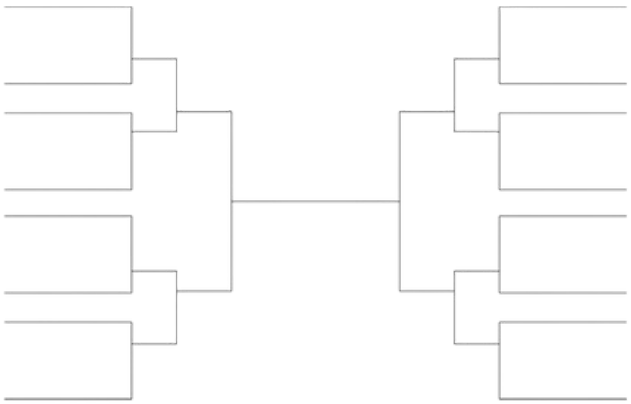
Stories submitted through *What's Your Story?* will help advance rare disease policy priorities through ongoing advocacy efforts to advance the equitable development of and access to lifesaving diagnoses, treatments, and cures. Submissions may be shared during meetings with Members of Congress or with the public via social media, online or in the press.

EVERYLIFE FOUNDATION FOR RARE DISEASES

FEBRUARY FUNDRAISING EVENT: MARCH MADNESS

March Madness Selection Sunday is March 14! Start assembling your pool and **raise funding for the resources and tools HCU Network America!**

March Madness is a three week period packed full of buzzer beaters and is a sports poolers dream. But how can you keep the excitement alive when most of your members have had their brackets busted? A great option is our Madness Squares pool format, as every game of the tournament will have a winner!



How Do Madness Squares Work?

If you are familiar with Super Bowl Squares, the main idea is the same for March Madness. A 10x10 grid of boxes is setup and each row and column is given a number from 0 to 9. Just like in Super Bowl Squares, each square of the grid can be claimed by a pool member.

Winner breakdown

Each round is worth a set number of points. You can determine this on your own, but be sure to let all of the entries know before the tournament begins what the scoring system will be. (You should write the point values under each round at the top of the bracket).

Declaring a Winner

Multiply the total number of correctly picked games in each round by the points assigned to that particular round. Tally all rounds together and the person with the highest point total wins!

- For further instructions and to print your bracket, visit: https://www.printyourbrackets.com/howtomarchmadness.html
- For online tools, check out: https://www.runyourpool.com/march-madness-squares-pools.cfm

NEED HELP HOSTING A FUNDRAISER? EMAIL US - INFO@HCUNETWORKAMERICA.ORG

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://hcunetworkamerica.org/contact-register/

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