The HCU *Herald*

HCU Hero
Juana from Uruguay

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

HCU HERO: JUANA FROM URUGUAY

Journey to Diagnosis

Juana was born in 2012 and was the perfect little baby. During her first years, everything looked well enough - she had some health problems but it wasn't enough to think there was an underlying condition. But I couldn't shake off the feeling that something wasn't right.

By age three Juana's vision was clearly impaired. On her first appointment with an ophthalmologist, it was determined that she already had severe myopia but it wasn't until age five that a different ophthalmologist identified movement in her lenses and said there was something else going on. We were told then that it could possibly be Marfan Syndrome.

After an internet search trying to learn more about Marfan Syndrome, I came across Homocystinuria as a possible alternate diagnosis, among many others. At that moment, I just felt that HCU was indeed what we were dealing with. I presented this new information to doctors but was met with resistance. Trying to convince them that it was worth testing her for Homocystinuria wasn't easy. It would be two years later that we got the actual official diagnosis: Classical HCU.

Even after her diagnosis was confirmed, getting the proper treatment was a whole new issue! Juana was the first patient in Uruguay to be diagnosed with HCU, and the medical formulas and medications that would be needed for her treatment simply aren't available here. Gaining access to these formulas and medications was a very long and difficult process.

What impacts did the diagnosis have on Juana and your family?

When we first learned about the treatment and protein-restricted diet, I felt as if life as we knew it was being taken away from us. On the one hand, we had answers to many things that had been going on with Juana, but on the other hand, I felt hopeless. So much had already been taken away from her and I feared that the dietary restrictions would only isolate her even more.

Because of her late diagnosis, Juana has had to miss out on many things. Her eye problems prevent her from being able to play most sports or even fully participate in gym classes at school. She struggles with her school work; not only her vision makes it harder but she has also been diagnosed with ADHD. She has a great relationship with other kids but she also has a hard time controlling her impulses, and this makes it hard for teachers to understand that she's not just spoiled, but that HCU has brought on other conditions which affect her behavior.

...so much had already been taken away from her and I feared that the dietary restrictions would only isolate her even more.

In the beginning, I tried to avoid restaurants and parties as it hurt too much seeing her looking at everything she had tasted before and couldn't have anymore. It was thanks to all the support from family and friends that we've received that now we both feel confident going out. Planning ahead and learning to cook and shop for the low protein diet has been challenging, but we're getting there.

Planning ahead and learning to cook and shop for the low protein diet has been challenging, but we're getting there.

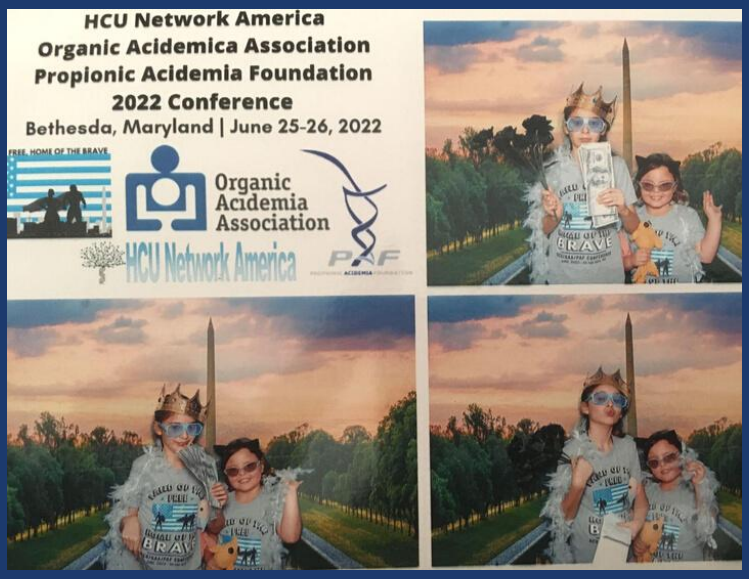
What doesn't cease to amaze me is how responsible Juana has been towards food restrictions, medication, and formula. She doesn't like her formula, but she knows she has to take it to be healthy, so she does. She never eats something she knows that she shouldn't, even if it's being offered to her.

What is the biggest challenge that you've faced?

I would say that the biggest challenge we've faced is the lack of information about HCU here in Uruguay. Luckily, I found HCU Network America and it has played a huge part in helping us to understand and to gain access to the treatments that Juana needed. Most doctors still don't know about Homocystinuria, so every time we go to a new doctor, I have to give a crash course in HCU first, which is not always appreciated. I long for the day when I can be just her mommy and not her primary care doctor too.

What have you been able to Celebrate?

This year we were given the opportunity to attend the HCU Network America conference in Bethesda, Maryland. It was a life-changing experience - meeting all of the families, and listening to the different experiences and information was just beyond words. Since the conference, Juana has said some things that made me realize just how important it was for her to meet other people with HCU. She's been asking me questions about treatment, clinical trials even, and what will happen in the future.



Juana & Samantha, having fun in the conference photo booth

She made new friends at the conference, and she felt welcomed. She told her uncle about her new friend Samantha, and it stuck with me that she said "She's got HCU just like me, and she's ok." As a nine-year-old, there are many thoughts and questions that she must have, with all the information she was told by me or even the things she's overheard in appointments. But it was only after the conference that she started to ask questions and talk about her concerns. She wanted to know if she would live past 30, (I don't know when she picked up that number), and I told her with her formula and diet she would live a great life and grow old and grey, so she reached the conclusion that the formula gave her a kind of superpower! She said to me "So with my formula, I'm going to be immortal!"

We have come SO far - her homocysteine levels have reached the normal range, which is the result of her effort and that was the greatest gift. I'm not so afraid of the future now, and I feel more hopeful after meeting other patients and familes.

I know I'll never stop advocating for Juana, I want to give her the best life possible just like any other mom wants.

What advice do you have for other families who may be coming off of a recent diagnosis?

Juana and her mom Anna, at the 2022 conference

For families going through the process of reaching a diagnosis, my advice would be: Trust your gut. YOU know your child, and if something feels off, keep pushing. For those who are receiving a diagnosis, look for support and find a community. Talking to people who are going through or have been through the same things makes a huge difference. Raising a child does take a village, and you'll be surprised to see how many people rally around you in support.

> ***I'm not so afraid of the future now, and I feel more hopeful after meeting other patients and families.***

This HCU Hero story was written by Anna, mom of Juana.

Jalapeño Popper Dip

***Yields ~12 servings* | Serving Size: 1 oz. (28g) | Protein per serving: 0.3 g**

Ingredients:

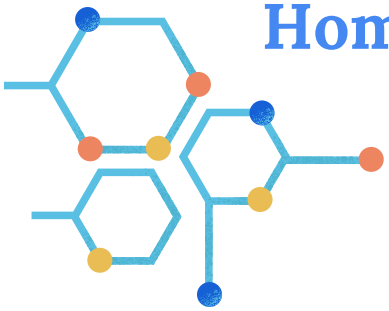
- 45 g raw jalapeño peppers (about 3 peppers)
- 134 g Violife Just Like Cream Cheese Original
- 1/2 c Mayonnaise
- 1 clove Garlic, minced
- 1/2 tsp Onion Powder
- 1/2 tsp Smoked Paprika
- 2 oz. Violife Just Like Cheddar Shreds
- 2 TBSP Sliced Green Onions
- 14 g Follow Your Heart Parmesan Style Shredded

Directions:

1. Turn the oven on to a low broil. Line a small baking sheet with foil and lightly spray with cooking spray. Place the jalapenos on the baking sheet and broil in the oven until roasted and blistered, making sure to keep an eye on them to prevent burning. Remove from oven and allow to cool a few minutes. Next, chop the roasted jalapenos and set them aside.
2. In a small saucepan combine the cream cheese, mayo, garlic, onion powder, smoked paprika, and roasted jalapenos. Place saucepan over medium heat, stirring occasionally. Heat until the cream cheese melts, and everything is well combined and smooth. Add the cheddar cheese shreds and green onions. Continue to heat until the cheese melts and the dip is smooth. Be sure to stir frequently to prevent burning. Remove from heat and put into a heat-safe bowl. Top with the parmesan shreds. Serve immediately.

INDUSTRY NEWS

Exciting things are happening in research for the Homocystinuria community!



Aeglea BioTherapeutics now has a clinical trial site open in the US for Pegtarviliase; a drug in development for the treatment of Homocystinuria due to Cystathionine Beta-Synthase (CBS) Deficiency. The site is located in Dallas, TX at UT Southwestern Medical Center and is led by Markey McNutt, II, MD.

To learn more, visit: https://bit.ly/3QYRajY

Aeglea BioTherapeutics Reports Second Quarter 2022 Financial Results and Provides Program Updates

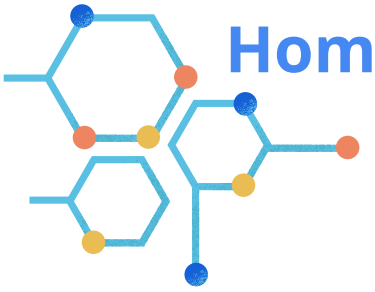
August 4, 2022 - Aeglea BioTherapeutics, Inc., a clinical-stage biotechnology company developing a new generation of human enzyme therapeutics as innovative solutions for rare metabolic diseases, today announced financial results for the second quarter ended June 30, 2022, and provided program updates.

"We continue to make progress with our Homocystinuria program and are encouraged by the physician and patient interest in advancing our Phase 1/2 trial. There remains a great need for better treatment options for Homocystinuria patients and we look forward to sharing data from this trial later this year," said Anthony Quinn, M.B., Ch.B., Ph. D., president and chief executive officer of Aeglea.

To read the full press release, visit https://bit.ly/3T9fMIv

INDUSTRY NEWS

Exciting things are happening in research for the Homocystinuria community!



Synlogic Granted FDA Fast Track Designation for SYNB1353 for the Treatment of Homocystinuria

August 23, 2022 - Synlogic, Inc., a clinical-stage biotechnology company developing medicines for metabolic and immunological diseases through its proprietary approach to synthetic biology, announced that the U.S. Food and Drug Administration (FDA) has granted Fast Track designation to SYNB1353 for the potential treatment of homocystinuria (HCU).

The FDA's Fast Track process is designed to facilitate the development and expedite the review of drugs to treat serious conditions and fill an unmet medical need. To qualify, available clinical and non-clinical data need to demonstrate meaningful therapeutic potential. The benefits of Fast Track designation include opportunities for frequent meetings with the FDA to discuss trial design, development plans and data needed to support drug approval, as well as the ability to submit a registrational filing for approval on a rolling basis, and eligibility for priority review, if relevant criteria are met. SYNB1353, an orally-administered, non-systemically absorbed drug candidate designed to consume methionine in order to lower homocysteine levels, is currently being evaluated in a Phase 1 study in healthy volunteers.

To read the full press release, visit https://bit.ly/3R6NcpI

Travere Therapeutics Provides Regulatory Updates on its Development Programs

Aug. 03, 2022 -- Travere Therapeutics, Inc. provided regulatory updates for its sparsentan programs in IgA nephropathy (IgAN) and focal segmental glomerulosclerosis (FSGS), as well as its pegtibatinase program for classical homocystinuria (HCU).

During their presentation, Travere announced the FDA recently granted Breakthrough Therapy Designation to pegtibatinase, the Company's novel investigational enzyme replacement therapy being evaluated for the treatment of HCU.

To read the full press release, visit https://bit.ly/3wu1bxL

HCU IN THE NEWS!

Last month, our very own Brittany and Grayson Parke were featured in Colorado's 5280 magazine!

FEATURES

Inside Colorado's Battle Against Rare Diseases

One in 10 Centennial Staters has a so-called orphan disease—the local medical community is trying to help them.

Barbara Urzua
5280 August 2022

Brittany Parke with three-year-old Grayson and a photo of her late infant son, Drew. Photo by Daniel J. Brenner

In 2011, doctors told Brittany Parke that her newborn baby boy was completely healthy, but she wasn't so sure. The 25-year-old was living in upstate New York, and as a second-time mom, she knew something wasn't quite right with little Drew. The two-month-old was sleeping excessively and refused to eat. Doctors brushed away her concerns. Then came the seizures. Three and a half weeks after first seeing a doctor, she began to receive answers. But by then, it was too late. Drew died shortly thereafter.

Genetic testing done on Parke and her husband offered an explanation: The pair had genetic variations that can cause a defect known as Cobalamin G disorder. The ailment brings with it poor feeding, slow growth, seizures, neurological disorders, and, if left untreated, death. It's not surprising Parke's New York doctors were stumped; the disease affects only one in 100,000 newborns.

Rare diseases such as Cobalamin G, often called orphan diseases, are conditions that each affect fewer than 200,000 people in the United States. Living with one can be a nightmare. Symptoms can be life-altering and life-threatening. Doctors often don't know what's wrong. Dealing with health insurance companies becomes a full-time job. And treatments are often as rare as the diseases themselves.

Colorado organizations are working to change that. In November 2021, the University of Colorado Anschutz Medical Campus—which includes UCHealth University of Colorado Hospital and Children's Hospital Colorado (CHC)—was designated a Rare Disease Center of Excellence by the National Organization for Rare Disorders (NORD), the nation's leading advocacy organization on rare diseases. CU Anschutz is the only medical center in Colorado to earn the title, but it is not alone in its mission to better the lives of those affected. Thirty other institutions nationwide received the designation when the program was launched last November and will collaborate to streamline the referral process. "We've been doing rare disease work and research for decades, although it's been happening in a way that's not very structured across the campus," says Dr. Matthew Taylor, director of adult clinical genetics at the CU School of Medicine. "Now that we have this center designation, we'll be able to organize all of our rare disease services and be in a better position for patients to connect with us more easily."

Check out the full article here!

https://www.5280.com/2022/08/inside-colorados-battle-against-rare-diseases/

HCU IN THE NEWS!

NIH Fellows Makes Plushies for Patients

These handmade plushies were handed out at our *Land of the Free, Home of the Brave* conference in Bethesda, MD!

TALENT BEYOND THE BENCH

NIH Fellows Make Plushies for Patients

Two fellows in Dr. Charles Venditti's lab spent many hours over several months crocheting what they're calling Pam and Pauline's Plushies for Patients.

"These are the coolest handmade stuffed animals I have ever seen," said Venditti, a senior investigator in NHGRI's Metabolic Medicine Branch.

Dr. Pam Head, an NIGMS postdoctoral research associate (PRAT) fellow, and Pauline Hoffman, an NHGRI postbac IRTA fellow, made 35 unique plushies for children who were stuck indoors at a medical conference in Bethesda over a weekend in June. The conference was for families grappling with MMA (methylmalonic acidemia), PA (propionic acidemia) and cobalamin deficiencies—rare, genetic metabolic disorders.

Head began crocheting in graduate school to pass the time during lectures. She started making stuffed animals earlier this year as gifts for colleagues returning to the office. When NHGRI clinical investigator Dr. Irini Manoli saw them, she asked whether Head would share a couple with patients visiting the Clinical Center. When she did, the patients' eyes lit up, recalled Manoli. "It was most heart-warming," she said.

With the summer family conference in mind, Head then teamed up with Hoffman and the two set to work during off-hours—on weekday evenings and weekends—to prepare one for each child attending the meeting.

"I became not just Pauline's mentor in the lab but also in crocheting," said Head.

"It was such a special set of gifts," said Venditti, who has devoted his career to studying organic acidemias. "All the kids got to pick one and even some of the unaffected siblings got plushies as well."

MMA and PA are life-threatening conditions that cause frequent hospitalizations and long-term complications such as learning and vision problems. While cobalamin (vitamin B12) injections can help milder forms, treatment largely consists of managing symptoms and, for some patients, organ transplants. Venditti's team focuses on developing genomic therapies for these disorders.

At the conference, the parents appreciated the plushies that brought their children joy and served the dual purpose of keeping their kids occupied so they could focus on the lectures.

"I hope the person who made them knows how happy the kids were to receive them," said Misty Garcia, whose daughter received a llama plushie.

Head and Hoffman said they hope to make more plushies in the future to donate to other young patients.—**Dana Talesnik**

Above, Grayson Parke, age 3, snuggles with his snake plushie. Below, Stephanie Evans poses with Dr. Charles Venditti and the dog and duck plushies she received. "She would not put them down," said Venditti." Same goes for the other kids.

At left, NIGMS postdoc fellow Dr. Pam Head poses with the plushies she crocheted for young people with MMA, PA and other cobalamin disorders. Sacha George (c), 8, who has cobalamin C deficiency, hugs his plushie. At right, Pauline Hoffman, an NHGRI postbac IRTA fellow, holds up a plushie she crocheted.

See full publication here: https://bit.ly/3ReIRAz

BREAKING NEWS!

HCU Network America Announces the Recipient of their First Newborn Screening Research Grant.

August 31, 2022 - The New England Newborn Screening Program, an initiative of UMass Chan Medical School's Commonwealth Medicine division, received the award to explore the development of reference ranges for additional newborn screening markers for early detection of classical homocystinuria and remethylation disorders. The research, led by Devinder Kaur, PhD, assistant professor of pediatrics at UMass Chan, aims to establish normal reference ranges for total homocysteine, along with other analytes collected by healthy newborns during the 24-48 newborn screening period. This will support the development of algorithms that will incorporate information on a variety of other variables in the future. Dr. Kaur, who is leading the research, joined the New England Newborn Screening Program in 2017 as a senior scientist.

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

Meet Dr. Kaur

Dr. Kaur is an assistant professor in the department of pediatrics at the UMass Chan Medical School. She received her master's and PhD from the Post Graduate Institute of Medical Education and Research, Chandigarh, India. She was a Research Scientist at the Colorado state University, Fort Collins, Colorado where she used an integrated approach of biochemistry, bioinformatics and genetics to decipher biosynthetic pathways of the mycobacterial cell wall with a view to new drug development and vaccine candidates for the treatment of tuberculosis. At Massachusetts Supranational TB Lab, she led activities ranging from providing technical assistance and technology transfer to developing countries, designing and implementing quality assurance systems, and drug potency testing, as well as practical and didactic training in diagnostic methods and quality assurance. She also participated in several operational research projects with academic research institutions and molecular diagnostic companies to identify mechanisms of drug resistance and to validate new diagnostic tests. She authored and co-authored articles in peer reviewed journals, on basic and operational research in the area of infectious diseases and newborn screening.

Dr. Kaur joined the New England Newborn Screening Program (NENSP) in 2017 as a senior scientist. She has served several roles and made strong contributions to various scientific and administrative operations of the program including the validation and implementation of a new Laboratory Information Management System, the program's short term follow-up responsibilities, optimization and implementation of a Covid antibody assay for a special collaborative epidemiological project and to the day-to-day clinical laboratory QC and troubleshooting activities of the metabolic laboratory. She is now leading the design, development and validation of improved and new mass spectrometry-based assays.

WALK, RUN, RIDE VIRTUAL RACE BENEFITING HCU NETWORK AMERICA

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?

- We rely on the honor system. You don't have to use a device to prove your miles.
- If you'd prefer to use an app to track your miles, we recommend Strava. You can join the HCU Network America Club.
- Please use intentional miles - this means no step counting
- Please log all your miles by 11:59 pm ET September, 30, 2022

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.

Learn more or register at https://runsignup.com/Race/IL/Batavia/HCURaceforResearch

info@hcunetworkamerica.org | https://hcunetworkamerica.org | 630-360-2087

SEPTEMBER 1-30, 2022

More swag available for fundraisers!

Per Individual: $30

Per Family (up to 4 – 1 mailing address): $50

Registration ends September 2!

https://runsignup.com/Race/IL/Batavia/HCURaceforResearch

Meet our Teams / Click to read each team's story & donate!

Brooke's Blazers	https://bit.ly/3dXWki8
Cakes for Carson	https://bit.ly/3PHGYLz
Carter Crew 4 HCU!	https://bit.ly/3QOqumo
Ellie's Entourage	https://bit.ly/3PRir6W
Grayson's Gang	https://bit.ly/3R5V4lt
Kristin's Krew	https://bit.ly/3wu7y4d
Masen's Movement!	https://bit.ly/3PJ7EM4
Mighty Marchese's	https://bit.ly/3R8qiya
Mississippi Madness	https://bit.ly/3PQQ4G7
Run with Shu	https://bit.ly/3wvr00h
Sylvia's Supporters - Recordati	https://bit.ly/3CN3OPp
Synlogic	https://bit.ly/3CyH1GU
Team Aeglea	https://bit.ly/3PJABHK
Team CbIG - Carson Hunt	https://bit.ly/3TgaHhP
Team Codexis	https://bit.ly/3pMwcco
Team Crows	https://bit.ly/3dQue8h
Team Recordati	https://bit.ly/3TgeCew
The Bartke Ruff Ruffs	https://bit.ly/3KiQwvq

Follow each team's progress throughout the month by visiting our race website!

https://runsignup.com/Race/IL/Batavia/HCURaceforResearch

Cobalamin C Research Fund $10,000 Match!

A very generous family has agreed to match donations made to the Cobalamin G Research Fund in the month of September during our Race for Research!!! (Up to $10,000)

Click here to donate!

Donate to Grayson's Gang, Team Carson Hunt OR create your own CbIG fundraiser! (In order to track and match donations)

Please share with your family and friends

Grayson's Gang

Team Carson Hunt

HCU NETWORK AMERICA

EVENT ANNOUCEMENT

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

FACEBOOK LIVE CHAT

Danae', Patient

September 6 at 8 pm EDT

Bone, Cartilage and Connective Tissue Survey

with the Homocystinurias

HCU NETWORK AMERICA

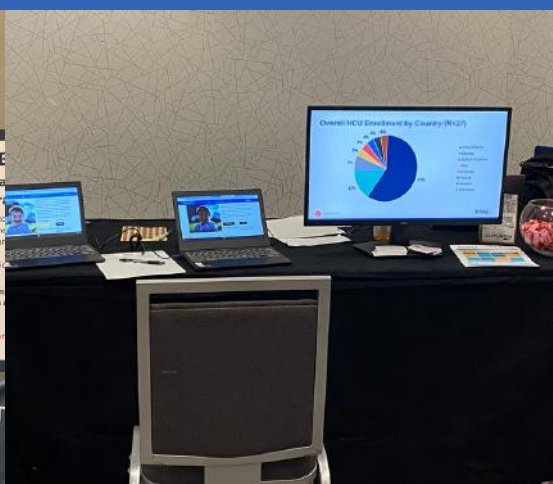
Tammi, Parent

Lindsey, Patient

Karen, Parent

Not a Facebook user? No problem!

You can register to watch here: https://bit.ly/3Aof55y



HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

Bone, Cartilage and Connective Tissue Survey

"...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. "

- Andrew, Classical Homocystinuria patient

Complete the Survey

homocystinuria.rare-x.org

HCU NETWORK AMERICA

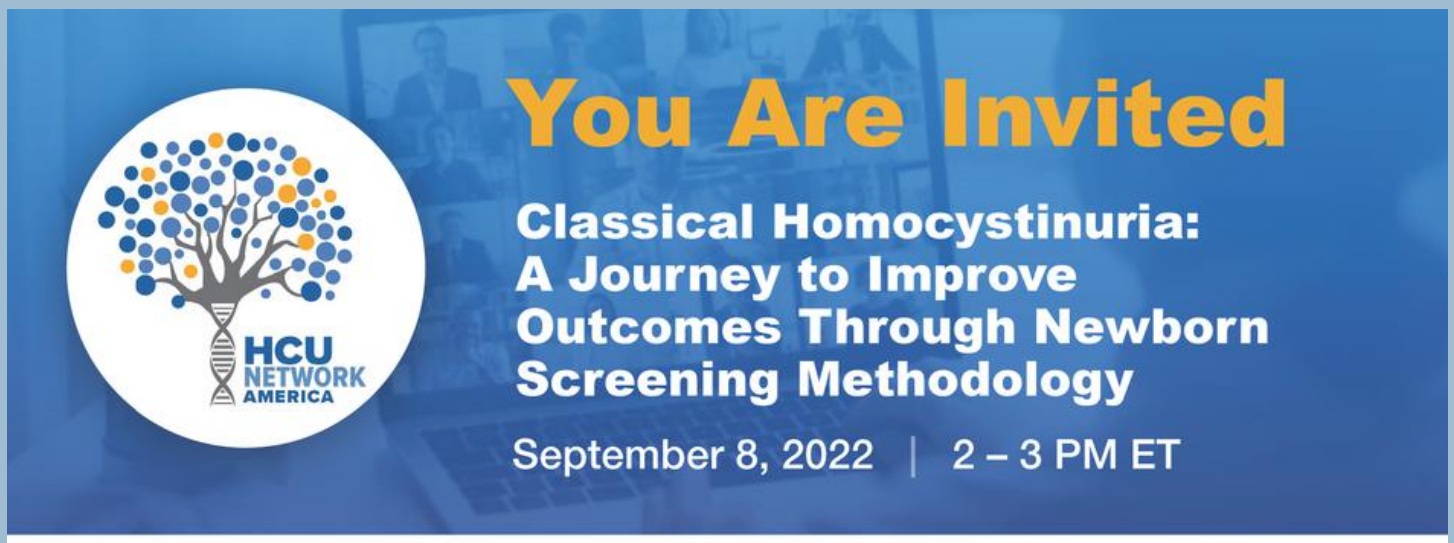
HCU Network Australia

POWERED BY RARE X

EVENT ANNOUCEMENT

September is Newborn Screening Awareness Month!

Join us for a deep dive into Newborn screening for Homocystinuria and how we can work together to support improved outcomes!



Data suggests more than 50% of newborns with Classical Homocystinuria (HCU) are missed by the screening methods used most commonly across the United States

Please join us Thursday, September 8th from 2 - 3 PM ET for a webinar to discuss:

- Current newborn screening landscape for Classical Homocystinuria (HCU)
- Impacts of delayed diagnosis on patients
- Recommended actions to improve newborn diagnosis rates
- How we can work together to support improved outcomes

Register here: https://bit.ly/3zxTS8O

EVENT ANNOUCEMENT

Calling all advocates!

Free Webinar!

Share Your Story Through Local Media

Interactive Webinar

Discover how local media can amplify your advocacy strategy

Britta Dornan,
EveryLife Foundation for Rare Diseases

September 8th, 2022
12:00 PM EDT

EVERYLIFE FOUNDATION FOR RARE DISEASES

RDLA Rare Disease Legislative Advocates
POWERED BY THE EVERYLIFE FOUNDATION

- ✓ Discover how local media can amplify your advocacy strategy
- ✓ Develop your news pitch for local reporters
- ✓ Find out how to write a news release
- ✓ Learn how to leverage local media coverage

Register here: https://bit.ly/3TaYlHR

UPCOMING EVENTS

Find all events at: https://www.eventbrite.com/o/hcu-network-america-30163980100

Classical HCU Parent-Caregiver Meetup

Parents,Grandparents and Caregivers of "kids" all ages with HCU need support too!
Saturday October 8, 2022
10 am CST / 11 am EST

Classical HCU Patient Virtual Meet-up

Online meet-ups are an opportunity to for patients impacted by homocystinuria to connect one another virtually.

Sunday, October 9 2022
2 pm CST / 3 pm EST

Cobalamin Disorders with HCU Community Virtual Meet-up

Online meet-ups are an opportunity to connect patients and caregivers impacted by cobalamin disorders with elevations of homocysteine to one another virtually.

Sunday, October 23 2022
2 pm CST / 3 pm EST

GET READY FOR HCU AWARENESS MONTH!

GET READY FOR HCU AWARENESS MONTH!

WEAR YOUR AWARENESS

October is HCU Awareness Month!
Get your Gear NOW!

https://www.bonfire.com/store/hcu-haberdashery/

Awareness Month Activities

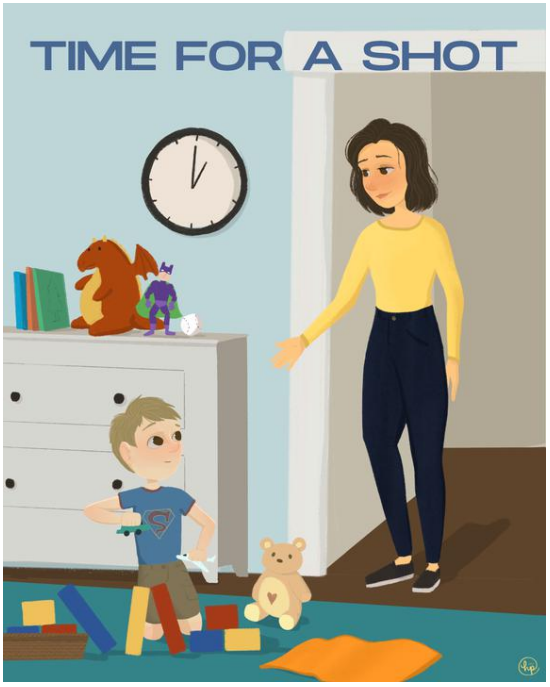
- ☐ Change your social media picture to the HCU Awareness Ribbon
- ☐ Start a HCU fundraiser
- ☐ Share an infographic about HCU
- ☐ Share a patient story
- ☐ Share your diagnosis story
- ☐ Challenge your friends to the same amount of protein and three normal protein shakes a day #ToastTocHCU
- ☐ Share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

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

NEW/UPDATED RESOURCE

TIME FOR A SHOT - A SOCIAL STORY FOR COBALAMIN DISORDERS

¡Ahora disponible en español!



Navigating a rare disorder is full of challenges, but you don't need to do it alone. As a community of families with patients diagnosed with Homocystinuria Cobalamin defects, our goal is to support and provide resources to help you care for yourself and your child.
Giving a young child a shot can cause a lot of stress and anxiety, for all involved. We hope this social story will help provide an opportunity for you to teach your child why they need to get a shot, how to ease the process, and educate them on the importance of medication management.

Download the full version of "Time for a Shot":
https://bit.ly/2YEsQhj

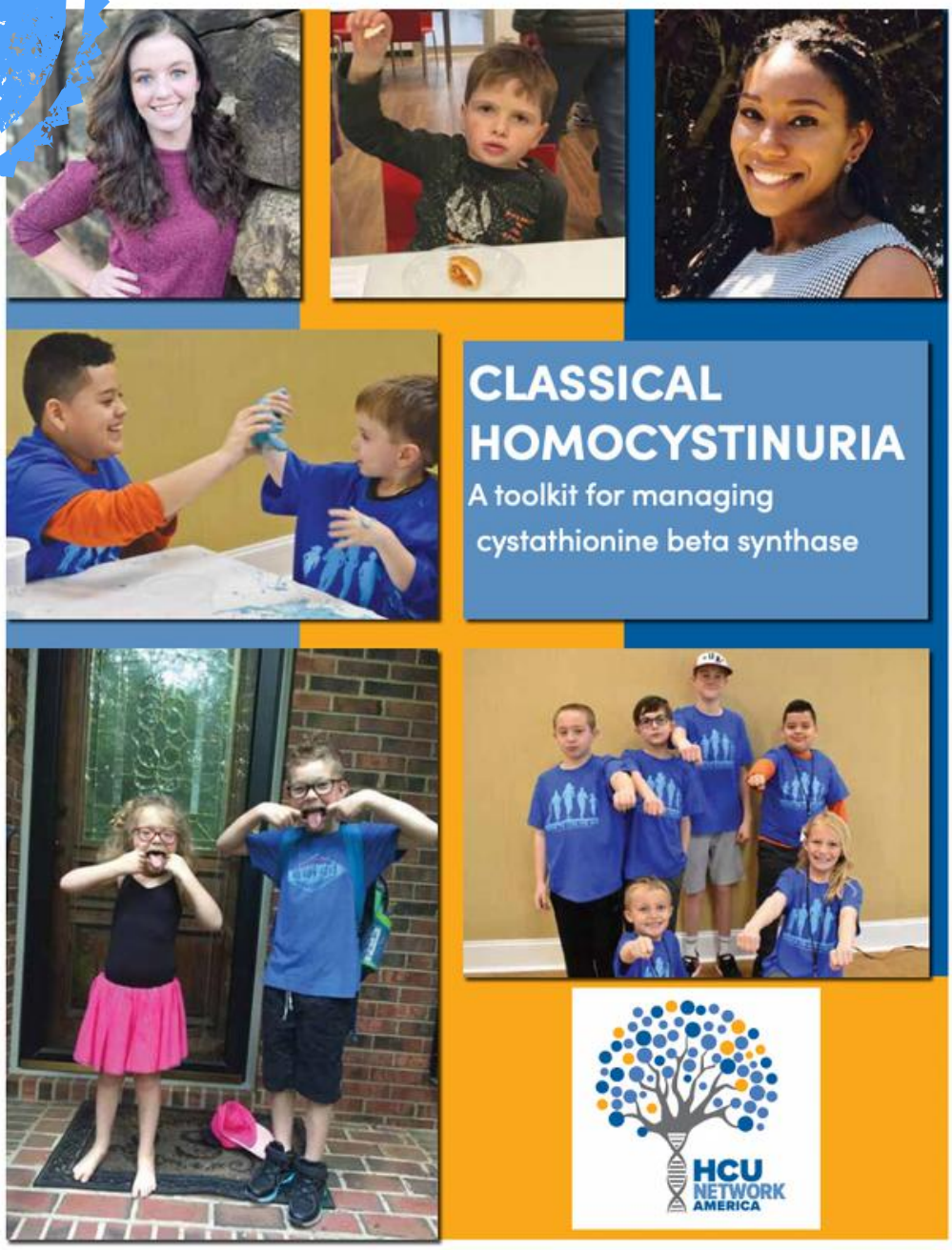
Navegar por un trastorno raro está lleno de desafíos; pero no es necesario que lo haga solo. Como comunidad de familias con pacientes diagnosticados con defectos de homocistinuria cobalamina, nuestro objetivo es apoyar y dar recursos para ayudarlo a cuidarse a sí mismo y a su hijo/a.
Ponerle una inyección a un niño pequeño puede causar mucho estrés y ansiedad a todos los involucrados. Esperamos que este cuento le dé la oportunidad de enseñarle a su hijo/a por qué es necesaria una inyección, cómo facilitar el proceso y como educarlo sobre la importancia del manejo de medicamentos.

Descarga la versión completa de "Tiempo para mi inyección":
https://bit.ly/3Kg4MFk

NEW/UPDATED RESOURCE

Check out our new look!

CLASSICAL HCU TOOLKIT



The Classical HCU Toolkit is designed to give you an overview of what Homocystinuria is, how it's treated, and how to better manage the diet and disorder.

Download the Toolkit:
https://bit.ly/3cvlZhs

MEET OUR NEW COMMITTEE MEMBERS!

MEET OUR CLASSICAL HCU STEERING COMMITTEE MEMBER

MELANIE

Melanie is the mom to Masen, an energetic 9-year-old little boy, who was diagnosed with Homocystinuria in June 2021 at the age of 8 years old. Masen was missed at newborn screening. It was at a routine eye doctor's appointment in February of 2021 where a very perceptive eye doctor noticed something going on with the lenses in Masen's eyes. After diagnosis, Melanie and her husband, Ryan were devastated and terrified at what the future held and struggled immensely while waiting to find out what damage had been done with so many years going undiagnosed. Masen underwent two separate eye surgeries (one for each eye) in April of 2021 to remove the lenses in both of his eyes. He now wears contact lenses and is seeing wonderfully with those.

Upon diagnosis in June 2021, Melanie didn't hesitate to jump right into researching and finding connections with others. After finding HCU Network America, Melanie was able to gain helpful information on how to move forward and let go of some of the fear and has made lifelong irreplaceable friendships with others in this extremely supportive community. Melanie hopes to be that same support to others while being actively involved in raising awareness for HCU.

MEET OUR CLASSICAL HCU STEERING COMMITTEE MEMBER

JAMELA

Jamela is a patient advocate and Registered Nurse, in South Florida. She was born in Brooklyn, New York where she was diagnosed with Homocystinuria through the Newborn Screening program. Since her diagnosis, she has been fortunate to have the support of her family and team of healthcare providers. The dedication of her family and admiration of her healthcare providers have made a significant impact on the trajectory of her life. Inspired by her community of care, Jamela hopes to be that type of advocate for others. She understands that patients can live optimally with the right type of resources and interventions. Jamela is excited to use her experiences as a patient and provider to bring awareness about the complexities of rare diseases and help create resources for the community. Living with a rare disease is not easy, but she hopes her story can bring hope to families facing similar challenges. Jamela currently serves on HCU Network America's Classical HCU Steering Committee and is a patient advocate with the Rare Disease Legislative Advocates (RDLA) through the EveryLife Foundation.

MEET OUR FUNDRAISING COMMITTEE MEMBER

DANA

Dana Hunt, who is a member of the Cobalamin Steering Committee, has joined the Fundraising Committee! Dana lives in the Kansas City suburbs of Kansas with her husband, Darren, and their two children, Ethan and Carson who has CblG. She loves being a mom, teaching dance, and spending quality time with her family. While this journey has been difficult, she reminds herself to live, laugh & love every day.

Compose

COMPOSE Study

A study that looks at the safety of pegtibatinase and how well it may work 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 may work in people with HCU at different dosage levels.

Approximately 32 patients will participate in sites in the US. The study, including a screening period, double-blind treatment period and an open-label extension (up to 138 weeks) 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 patients who participate in this study.
To inquire about participation in the study, **please contact:**

HCUConnect@labcorp.com

Visit **www.hcuconnection.com** for more information.

TRAVERE™ THERAPEUTICS

MA-PE-22-0002. February 2022

NOW ENROLLING:
A Natural History Study on Classical Homocystinuria

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

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

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

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

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

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

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

If you have any questions, please email:

HCUConnect@labcorp.com

Visit www.hcuconnection.com for more information

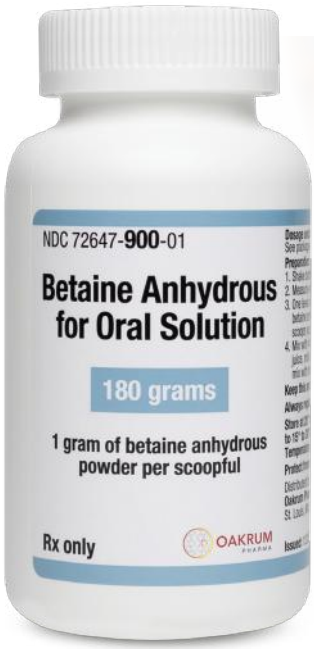
MA-PE-22-0004. March 2022.

TRAVERE™ THERAPEUTICS

NOW AVAILABLE

Betaine Anhydrous for Oral Solution

*The **FIRST AB RATED GENERIC** Version of*
CYSTADANE® (betaine anhydrous for oral solution) Powder



NDC NUMBER	STRENGTH	FORM	TE RATING	COMPARE TO
72647-900-01	1 gm/scoopful	Powder	AB	CYSTADANE®

PRESCRIPTION ORDER FORM

PRESCRIPTION CO-PAY CARD

INDICATIONS AND USAGE

Betaine Anhydrous for Oral Solution is indicated for the treatment of homocystinuria to decrease elevated homocysteine blood concentrations in pediatric and adult patients. Included within the category of homocystinuria are:

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

IMPORTANT SAFETY INFORMATION

WARNINGS AND PRECAUTIONS

Hypermethioninemia in Patients with CBS Deficiency: Betaine Anhydrous for Oral Solution may worsen elevated plasma methionine concentrations and cerebral edema has been reported. Monitor plasma methionine concentrations in patients with CBS deficiency. Keep plasma methionine concentrations below 1,000 micromol/L through dietary modification and, if necessary, a reduction of Betaine dosage.

ADVERSE REACTIONS

Most common adverse reactions (> 2%) are: nausea and gastrointestinal distress, based on physician survey.

To report SUSPECTED ADVERSE REACTIONS, contact Oakrum Pharma, LLC at 1-833-444-8010 or FDA at 1-800-FDA-1088 or www.fda.gov/medwatch.

Click here for full prescribing information.

OAKRUM PHARMA

3636 S Geyer Road, Suite 100
St. Louis, MO 63127

It was great meeting all you HCU Heroes at the recent conference in Maryland! In case you were unable to attend, here is what Vitaflo offers the HCU community:

Our range of formulas provides a variety of options to fit the demands of a modern lifestyle.

Prepare a semi-solid consistency or a low volume drink!

HCU gel™

- ✓ Suitable from 1 year of age
- ✓ Pre-measured powdered formulas containing 10 g protein equivalent (PE) per packet
- ✓ Mix with a small amount of fluid to be taken as a spoon feed or a drink
- ✓ Unflavored

Provide flexibility to prepare a variety of delicious drink options.

HCU express plus™15
HCU express plus™20

- ✓ Suitable from 3 years of age
- ✓ Pre-measured powdered formulas containing 15 g or 20 g PE per packet
- ✓ Mix with approximately 3 fl oz water or other permitted beverages to a low volume drink
- ✓ Unflavored

Ready-to-drink options on-the-go to school, work, or travel.

HCU cooler®15

- ✓ Suitable from 3 years of age
- ✓ Ready-to-drink formulas containing 15 g PE per pouch
- ✓ Available in Red and Orange flavors

For our collection of low protein recipes, check out the recipes section on our website **VitafloUSA.com/recipes**

For more information about Homocystinuria (HCU) products and to request a sample, visit **VitafloUSA.com | Vitaflo.Ca**

FOR USE UNDER MEDICAL SUPERVISION

Be sure to check with your healthcare professional before making changes to your diet.

FOR REIMBURSEMENT ASSISTANCE, VISIT:

formula4success®

Vitaflo™

Enhancing Lives Together

A Nestlé Health Science Company

Live better, together!

Contact Register

What is the contact register?

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

What will this information be used for?

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

How do I participate?

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