The HCU *Herald*

HCU Network America

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

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









COBALAMIN STEERING COMMITTEE

Representing Cbl C, F, G -with hopes of E, J and X to join us!

JOIN US!

Help drive the future of COBALAMIN DISORDERS

LEARN MORE ABOUT THE COMMITTEE AT:
http://bit.ly/HCUNAChISC_OR

HCU HERO: HENRY FROM MISSOURI

Henry was born in 2020 in Missouri. I had a normal pregnancy and delivery. He was born at 39 weeks, but only weighed 5lbs 13oz. This was my first clue that something was up. We struggled to keep his sugars where they needed to be after delivery, but they assured me this was common in low weight babies and not to worry. We went home a couple of days later.

The day after we got home, I got a call from a genetic counselor telling me Henry had markers show up on his newborn screening. They couldn't tell me if anything was definitely wrong or not, but they did say that if something was wrong, it could potentially be very serious. They told me to feed him every 3 hours, and if he started to not tolerate his feeds then to take him to an ER. The only thing I noticed was he did seem to sleep a lot, but he was a newborn so I didn't think too much of it. We ended up having blood drawn and starting another medication while we waited for some results. We also ended up doing a B12 injection 1-2 times a week until we got the results back; which seemed to take forever. We finally were told that it was Cobalamin C (CblC) and that Henry would have to eat on a strict schedule and that he would have to take lots of medications, including a daily shot of B12 that they trained us to do.

This was an incredibly difficult time. Waiting was the worst; not knowing what was wrong, let alone how to fix it and having this TINY baby to take care of. Then having to go through all the blood draws and having to do shots - it's so hard to watch your little one go through all that. Being the one to actually give the shots was hard too; I was the one hurting him.

Once we got him on all the meds he needed, we finally were able to start trying to settle in and form a routine. Henry grew well and met all of his milestones. He is a ridiculously happy child who is absolutely bursting with personality. He's always been very alert, even as a newborn. When he was awake he was very focused on you. He runs around with his older siblings all the time and is very bossy with them. Being outside is definitely his favorite. So far Henry has been asymptomatic and with the meds, his levels are well controlled. He's a little delayed in his speech, but it's hard to say if that's due to his disease or just because he's the baby and we all cater to him.

Having a child with a rare disease is incredibly hard. The unknowns, the worries, the what ifs, the treatments and schedules, the appointments and lab draws, and medical bills. It's a lot, and it's hard to watch him go through it all. But it is also so much more than worth it. Henry is thriving and he enjoys life every day, even on the hard ones. We are so grateful that God gave us this beautiful boy.

Email us your patient story! info@hcunetworkamerica.org

15 PE HOMACTIN™ AA

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LEMON LIME

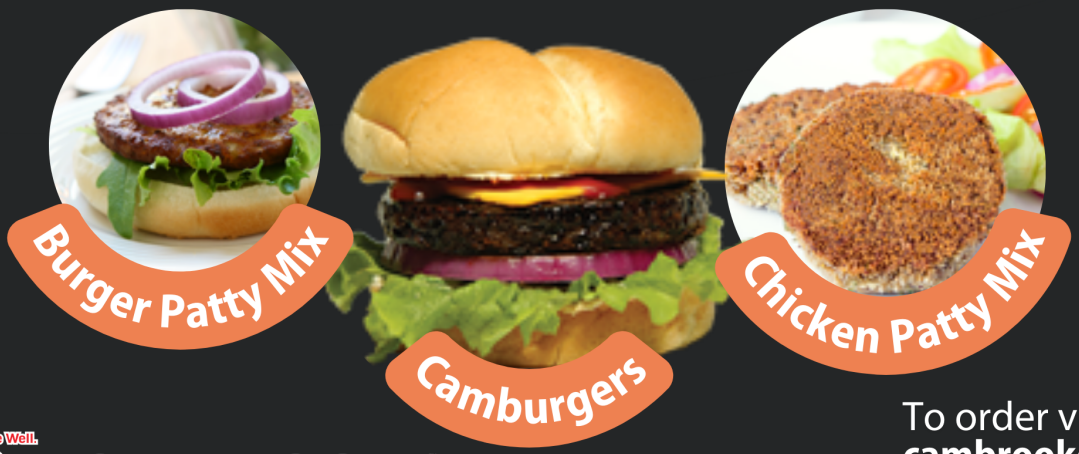
FLEXIBLE
15g PE & 150 kcal

LOW VOLUME
5oz WATER

OPTIMIZED
BONE HEALTH PROFILE

samples.cambrooke.com

Try our versatile meat alternatives



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cambrooke.com
866 456 9776

Chile Verde Jackfruit

***Yields servings* | Serving Size: 40 g | Protein per serving: 0.5 g | Calories per serving: 23**

Ingredients:

- 1 20-oz. can Jackfruit, canned, drained
- 1/2 c Vegetable Broth
- 1/2 tsp Garlic Powder
- 1/2 tsp Onion Powder
- 1/2 tsp Smoked Paprika
- 1/4 c Enchilada Sauce, Green Chili
- 2 TBSP Sour Cream

Notes

- This can be used to make tacos, taco salad, nachos, burritos, and enchilads

Directions:

1. Rinse the jackfruit well and remove all seeds. Squeeze excess water from the jackfruit and set aside. Add 2 tablespoon of olive oil to a medium skillet. Heat the oil over medium heat. Once heated, add the jackfruit. Sauté until lightly browned.
2. Combine all other ingredients in the skillet with the jackfruit. Stir to combine and continue to cook until the jackfruit is tender. Remove from heat and pull apart with forks. Serve immediately.

SPEAK UP FOR THE MEDICAL NUTRITION EQUITY ACT

1. **The best way that you can help make sure that the Medical Nutrition Equity Act (MNEA) moves this Congress is by asking contacting your members of Congress NOW.** Please go to nutritionequity.org/contact-congress and use the simple form to send an email to your members of Congress asking that they co-sponsor the bill. It doesn't matter if you have sent an email before. We encourage them to keep asking your members of Congress to sign-on until they do so.
2. **Please also ask contribute stories to the website.** The patient story form is here and the one for medical providers is here. We currently have ZERO or very few stories from a few key states, including Maine, Rhode Island, Mississippi, and several others.
3. **If you have had a meeting with a congressional office in the last few months asking them to co-sponsor the bill, please make sure that you are following up with those offices to see if they have any questions, or what they need to become co-sponsors.** The coalition has created a webpage with a wide-range of resources that should answer most questions that come up, but if you don't see what you need, please let me know.

We can get MNEA passed if we all work together to build the support that we need!

The Patients & Providers for Medical Nutrition Equity Coalition

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: French Toast
Lunch: Spring Rolls
Dinner: Stuffed Pasta & Salad

T

Breakfast: Smoothie & Breakfast Bar
Lunch: Nuggets & Watermelon Salad
Dinner: Fajitas & Dessert

W

Breakfast: Bagel and Fruit
Lunch: Grilled Cheese
Dinner: Italian Pasta Salad

T

Breakfast: Blueberry Lemon Muffin & Yogurt
Lunch: Sweet Potato Carrot Soup & Fruit
Dinner: Beet burger & Coleslaw

F

Breakfast: Cereal & Berries
Lunch: Gazpacho & Torilla chips
Dinner: Burrito & Jicama Sticks

Shopping List

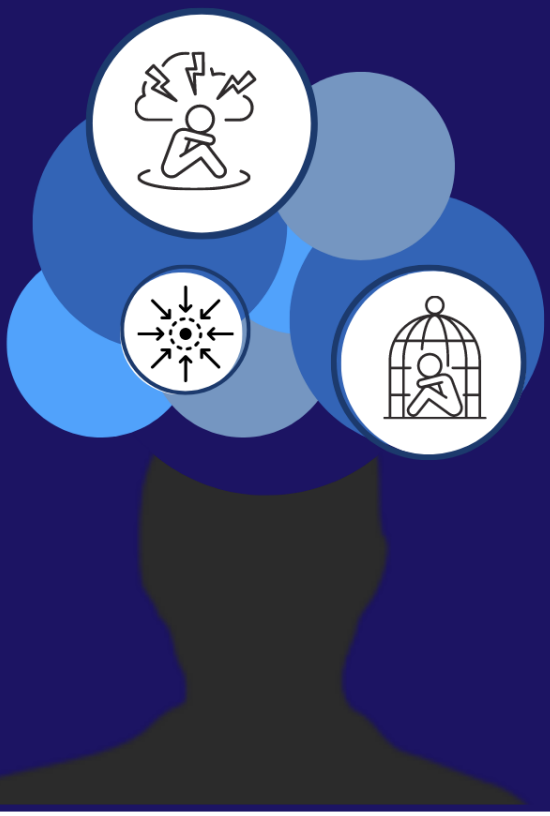
Click each day to view the week long menu!

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.

RESEARCH OPPORTUNITIES

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

In a study from a 2019 publication, "Revising the psychiatric phenotype of homocystinuria", 16 of the 25 patients in the sample (64%) reported psychiatric symptoms, including a high prevalence of both anxiety (32%) and depression (32%). Deficit–hyperactivity disorder (ADHD), oppositional defiant disorder (ODD), mood swings, hallucinations, and suicidal thoughts have also been reported.



Complete the Behavior Survey

homocystinuria.rare-x.org

HCU Network America HCU Network Australia POWERED BY RARE-X

UPCOMING EVENTS

HOMOCYSTINURIAS DATA COLLECTION PROGRAM

FACEBOOK LIVE CHAT

August 4 at 4 pM EDT

Behavior and Mental Health

with the Homocystinurias

Danae', Patient

HCU Network America

Brooklyn, Patient

Chrissy, Parent

Joanna, Patient

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!

August 20, 2022 at 10 am CT

Come network and learn from other parents, grandparents and caregivers!

Classical HCU Community Virtual Meet-up

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

Sunday, August 21, 2022 | 12 pm PT | 3 pm ET | 8 PM UTC

ICYMI RESEARCH NEWS

Exciting things are happening in research for the homocystinuria community!

Synlogic Initiates Phase 1 Study of SYNB1353 for the Treatment of Homocystinuria (HCU)

July 12, 2022 -- Synlogic, Inc. a clinical-stage biotechnology company developing medicines for metabolic and immunological diseases, announced that it had dosed its first healthy volunteer in its Phase 1 study of the investigational oral therapy SYNB1353 designed to consume methionine for the potential treatment of homocystinuria (HCU).

SYNB1353 is a candidate drug designed to provide a safe, orally administered, non-systemically absorbed treatment to consume methionine in the GI tract so it is not absorbed and converted to homocysteine. This hopefully will lower homocysteine levels in patients with Classical HCU, thereby lowering the risk of serious and debilitating complications. The company expects data from the SYNB1353 healthy volunteer study in the second half of 2022

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

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Drawing Blood to Advance New Therapies

Organizations that advocate for patients with rare diseases have a lot on their plates. Their leaders —individuals often affected by these diseases in some way—wear many hats. They become quick experts in genetics, fundraising, community building, and the needs of researchers working to better understand these disorders and develop new therapies.

When it comes to collecting biosamples for research—often one of the most important requirements of scientists—Coriell can help shoulder that burden.

Danaé Bartke is the executive director of HCU Network America, an advocacy group for individuals with homocystinuria (or HCU). The homocystinurias are a collection of heritable metabolic disorders that can cause serious symptoms affecting the eyes, brain, skeleton, and vascular system. The most common disorder in this group is called classic homocystinuria, the disorder with which Bartke herself has been diagnosed.

HCU Network America recently held their third annual family conference in Bethesda, Maryland, this year in cooperation with fellow advocacy groups Organic Acidemia Association and Propionic Acidemia Foundation.

One priority of HCU at this year's event: collecting blood samples from patients and affected family members so researchers have the biomaterials they need to develop therapies.

"One of the first questions we get from researchers is 'Do you have a biobank?'" Bartke said.

This is what led Bartke to Coriell and the National Institute for General Medical Sciences' Human Genetic Cell Repository. This collection of biosamples at Coriell contains cell lines and DNA representing a wide range of heritable diseases and is always working to add more.

"Our organization's president, Margie McGlynn, learned about the collection in a conversation with someone in leadership at Coriell," Bartke said. "We were thrilled to learn that so much of the cost is covered by the collection itself and we reached out to the team at Coriell right away."

At this year's event, a Coriell team working for the NIGMS Repository was on site to present information about the collection, the importance of building a biobank, and to answer any questions from families interested in donating. They also arranged everything needed so patients and their family members could have their blood drawn at their convenience. Biobanking can be a complicated and expensive initiative for advocacy groups. The NIGMS Repository makes is free for users to contribute and its samples are available to scientists around the world for a low cost.

"As a patient, you hear about repositories where researchers have to pay high prices for samples, and as a patient, we're giving part of ourselves to it. We want these samples to be available widely," Bartke said. "Coriell makes it affordable."

At Coriell's booth, families involved in HCU Network America as well as those present from the Organic Acidemia Association and Propionic Acidemia Foundation could have their blood drawn free of charge. For Bartke, a main organizer of the weekend's event that included families from as far away as the United Arab Emirates, this was one less thing off of her plate.

"The blood draw was one of the things I had to work on the least. Coriell did their own presentation, they had their own booth, they hired and brought their own phlebotomist," she said. "It's great that Coriell makes it so easy."

Her pitch to families interested in contributing was simple.

"You donate blood one time and you've contributed to this vast wealth and future that can improve things not just for yourself, but all patients," Bartke said. "If we're ever going to have a better understanding and develop better therapies for our diseases, we all need to be in this together. Not just researchers, not just the pharmaceutical industry bringing these therapies to market. It's everyone working together to make it happen.

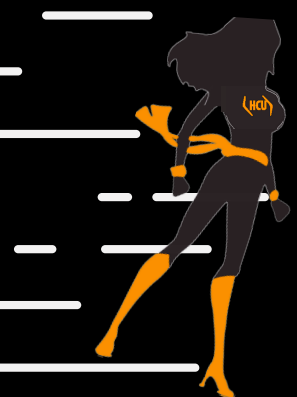
If you are a patient, or carrier and would like to participate, please reach out to NIGMS@coriell.org.

BE A HERO
HELP ACCELERATE RESEARCH

Cystadane; the last FDA approved drug for homocystinuria was approved in 1996. That was 26 years ago!

Currently there are three clinical trials in progress for classical homocystinuria, but there are none in progress for remethylation disorders.

In collaboration with other organizations, HCU Network America has issued 4 research grants. Each grant has been $40,000.



Help us accelerate research for better treatments for all homocystinurias and participate in raising funds during our Race for Research!

Register at: https://bit.ly/HCURace

SWAG INCLUDES

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

Are you the caregiver of someone with a chronic medical condition requiring support?

JOIN OUR RESEARCH STUDY!

The goal of our study is to understand how caregivers manage their lives when caring for someone with a chronic medical condition over time.

You may be able to participate if:

- You are 18 years or older
- You are the caregiver of a person with a chronic medical condition or you support someone who cares for a person with a chronic medical condition
- You are willing to share your experiences about caregiving

What will the study include?

- Annual online and phone assessments
- Periodic daily diary logs
- An optional blood sample
- A chance to invite other adults closely involved in your life to participate
- Compensation for your time

The study is completely voluntary and you may choose to stop participating at any time.

If you are interested in participating in the study or have any questions, please call us at **301-219-3394**
or email us at **CaregivingStudy@mail.nih.gov**

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

JOIN OUR FUNDRAISING TEAM

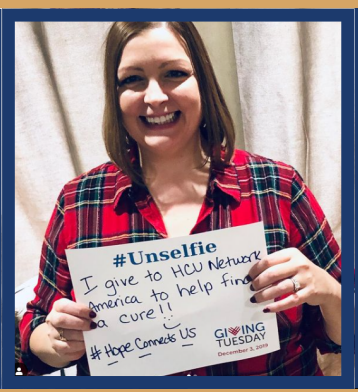
We are looking for new community members to join our fundraising team!

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

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

Email us:
dbartke@hcunetworkamerica.org

Meet our fundraising team!



Help us make future therapies a reality!

BIRTHDAYS

AUGUST BIRTHDAY?

Create a
BIRTHDAY FUNDRAISER
to help homocystinuria patients

HCU Network America

Create your own birthday fundraiser to raise money for HCU Network America. Go to https://www.facebook.com/fund/HCUNetworkAmerica/

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:

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Vitaflo™

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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