The HCU Herald

Featuring...

HCU Hero
Amy from New York

August 2023

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

HCU HERO:
AMY FROM NEW YORK

We are proud to introduce our daughter, Amy, to this group and hope that there might be some things that people read that can be useful in the treatment of their children. She was born on February 28th, 2008, thereby avoiding a potential Leap Year birthday. Now, as a 15-year-old about to go to the public high school that her older brother graduated from and where her older sister is going to be a senior, Amy is a tall, pretty, artistic, fashion-loving young woman, who is painfully shy at first but is by nature very sweet. Her face lights up when she hears that we are going to be in the presence of any of a number of babies in our family. Once she started being tested/examined by experts, she showed a toughness and bravery which she doesn't always give herself credit for, but that we remind her of constantly.

Unlike most of the other homocystinuria families that we have connected with, Amy did not receive her diagnosis of homocystinuria due to severe MTHFR deficiency until she was almost 15 years old. Unfortunately, there was no newborn screening done for severe MTHFR deficiency, which is not on the recommended list of diseases to screen for. Amy is the third child in our family, our baby, and her brother and sister are neurotypical, healthy teenagers (ages 17 and 19). My pregnancies were normal, except for gestational diabetes, which I had during all three pregnancies. The diabetes was well controlled by insulin, and the children were unaffected at birth.

Amy was a happy baby, who appeared to be on the same track in terms of developmental milestones until she reached about two years. When my mother expressed concern for her development, I made excuses by pointing out that Amy was often put in and out of the stroller/ car seat while we ran around to her siblings' daily activities, and rarely took a nap at home in the crib. Similarly, I attributed her delayed speech to having her needs met by her siblings' requests. After attending half day nursery school at the ages of two and three, she was recommended for early intervention speech services at the age of 4, which she received twice weekly until she was declassified going into kindergarten. Amy was happy in kindergarten, but her teacher expressed concerns about her ability to recognize basic numbers, letters, colors and we noticed that she struggled with coordination. She also struggled with her sense of time, and the ability to remember anything sequential: days of the week, months, or words to songs. In the first grade she was tested by the school district and was offered speech therapy twice a week. Amy also began to struggle socially, as friends and classmates naturally became more discriminating and as friends embraced and excelled at sports and dance activities that were too challenging for her. When we witnessed a group of her peers chatting at social gatherings, it seemed that the pace of their conversation was too fast and in time their interests grew further apart.

We applied to a specialty private school for language-based learning disabilities and had to do some private testing with a neuropsychologist for her application. One neurologist suggested that she might be having "mini-seizures" when it appeared that she was staring into space. She had an abnormal EEG in the office, showing micro-seizures when provoked, but a 72-hour video EEG was normal. She continued to walk with severe "in-toeing" but the pediatric orthopedic thought she would grow out of it. Similarly, her pediatrician was unconcerned.

Amy attended a private school from second grade through ninth grade which had extremely small class sizes, modified curriculum, and assessments. Amy had a lot of social anxiety and would get sick almost every morning before school. She took medication for ADD and the anti-anxiety medication which was very helpful and resolved her morning stomach issues. We continued to have her IEP done by the public school of record throughout her time at the private school, in case they thought she was ready to return to the public school so that she would at least receive the tri-annual testing. She was classified as Learning Disabled at this time.

In April of 2020, Amy had her first focal seizure while on a walk with her dad. She collapsed and was staring at the sky and was unresponsive to her dad's attempt to get her attention. After about 3-4 minutes, she came to, and he was able to support her enough to get her home. She was diagnosed with epilepsy and began taking Keppra to control the seizures. We also had her tested for autism, and she was diagnosed with that in 2021. A few months later she had another seizure during which only half of her body was seizing. There is some debate among her doctors as to whether this was actually a stroke. Around this time her unusual gate became more pronounced. In early 2022 we did genetic testing and learned about the Severe MTHFR deficiency. Her homocysteine level was over 200. It is better controlled now, with her numbers between 70-80.

However, in May of 2023, Amy told us that she was having trouble seeing and that she was seeing “dots”. After a visit to a retina specialist, we were told that her optic nerve was swollen and that we should take her to the ER. She ended up having a spinal tap to relieve the intracranial hypertension. Amy is now taking Diamox to prevent further buildup of excess fluid. It is hard to pinpoint what caused the excess cerebrospinal fluid; it could have been a side effect of the medications she takes, her genetic condition, or related to being overweight. She now also wears AFO braces on her feet, receives botox injections in her legs and hips and physical therapy to help with her gait disorder.

This spring Amy also received eligibility through the OPWDD (Office for People with Developmental Disabilities) and is currently attending her first camp program for individuals with disabilities. She will be in a similar program at our public high school this fall. Dropping her off at camp with teenagers and young adults with Downs Syndrome, autism and other physical and developmental conditions was a new experience for us. Given that her progressive condition and resulting cognitive decline has occurred mostly in the last few years, we didn’t know how she would feel about being with new people, some with more obvious physical challenges and some almost ten years older than she. After a month of camp, it seems like it has been a good experience for her. She talks about having friends, and for the first time in a long time, she describes being good at things, even helping other campers. While this makes us very happy, it also reminds us that it must have been painful for her to have struggled to keep up with others - physically and mentally - in a typical classroom for so long.

“While this makes us very happy, it also reminds us that it must have been painful for her to have struggled to keep up with others - physically and mentally - in a typical classroom for so long.”

We knew nothing about homocystinuria or Severe MTHFR deficiency. Most of her doctors don't know much about it either, and I'm not even sure if anyone does newborn screening for it in NY. For many years we treated her symptoms in isolation, having no idea that a genetic test could provide an answer for us. It is upsetting to think that if we had known to get her tested sooner, and if we had started treatment at a much younger age, she might be in a very different situation at this time. We have a letter from her geneticist that we carry with us in the event she is in an emergency room, as well as a list of all of her medications, so that we can explain her condition to the doctors on call.

It is hard to write about how having a child with special needs impacts the entire family, although the effects are considerable. It is particularly challenging given the fact that this could be read by any of our family members, and we don't want any one of us to feel culpable in any way. As parents, we know that having a child with special needs can be isolating, and we have experienced anger and frustration at our situation. We struggle to practice patience, with our children and ourselves, knowing that we are all experiencing a sense of loss. Our other children have had to be independent, and while ultimately this may benefit them in some ways, losing time with parents because they are busy with caretaking and the logistics that come with frequent therapies and doctors' appointments will no doubt have impacted them, as being independent and being forced to be independent are very different situations. Amy's sister, Kiera, suffers from AMPS (Amplified Musculoskeletal Pain Syndrome) which was also diagnosed through genetic testing, and fibromyalgia, which we want to mention in the event that there is a connection between the two genetic conditions. Having more than one child with medical issues is something that also appears to be common in families with metabolic genetic disorders.

Fortunately, we are blessed with family and friends, who help with driving to appointments, financial assistance (co-pays and parking in Manhattan where she has had more than 20 appointments in the past year adds up!) and many who have helped us by listening. Even though it must be irritating when our conversations with friends inevitably involve discussing our latest medical news, it helps us process what is happening and just listening helps more than they know.

We are so proud of Amy, as well as her siblings, Kiera and Brendan. If a hero is someone who combats adversity with strength, they have ALL shown remarkable heroism in the face of this challenging situation. Amy is our HCU hero because she demonstrates strength on a daily basis when she takes her medicine and suffers from its side effects without complaint. She has accepted her condition with grace and by example, teaches us to practice patience and inclusivity.

Did you know?
There's a support group on Facebook for families living with severe MTHFR.

Homocystinuria due to Severe MTHFR Deficiency Global Support Group

https://www.facebook.com/groups/353276269557147

To read Amy's story on our website, visit
https://hcunetworkamerica.org/amy-new-york/

Will YOU be our next HCU HERO?

Tell us:

- How you or your child was diagnosed?
- How has HCU affected you, your family and relationships?
- What are some of your successes with HCU
- What are some of your challenges you have faced?
  - How have you overcome them?
- What words of advice would you give to newly diagnosed families?
- For other patient stories, visit: https://hcunetworkamerica.org/patient-stories
- Email your story to: info@hcunetworkamerica.org

JOIN OUR FUNDRAISING TEAM

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

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

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

Email us:
dbartke@hcunetworkamerica.org

BIRTHDAYS

AUGUST BIRTHDAY?

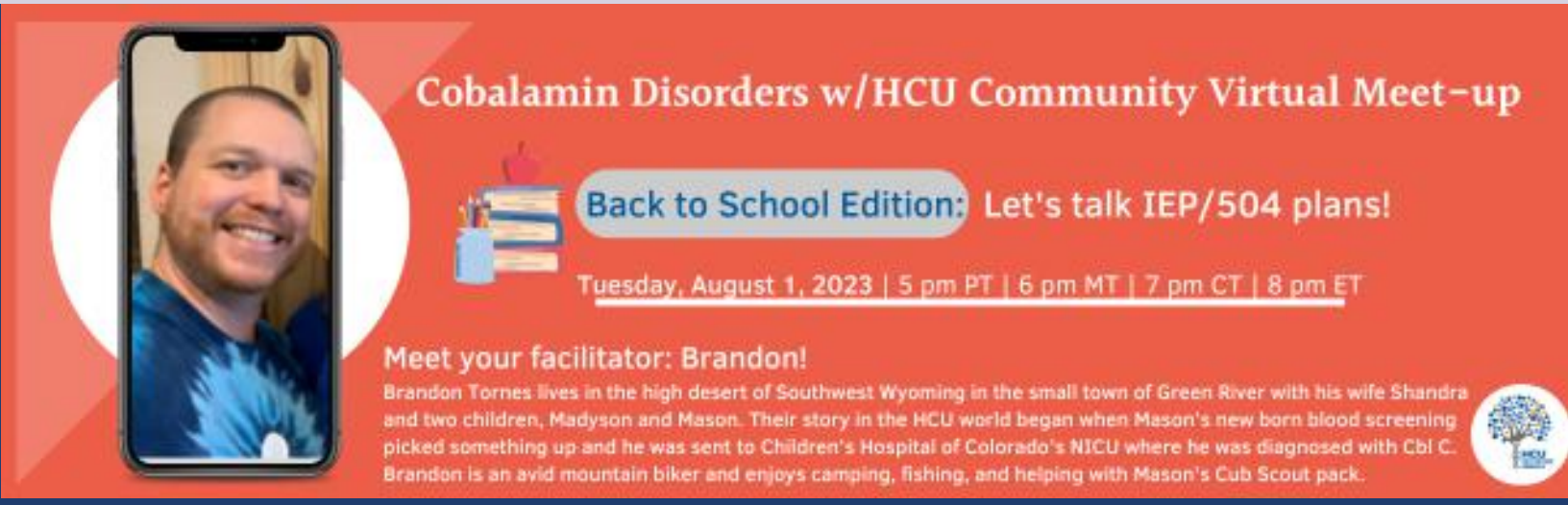
Create a
BIRTHDAY FUNDRAISER
to help homocystinuria patients

HCU NETWORK AMERICA

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

UPCOMING EVENTS

Register for these events at: https://hcunetworkamerica.org/virtual-meet-ups/



What should I expect?

Come join us on September 28 at 4 pm ET

Hear from Larry Bauer and James Valentine, from Hyman, Phelps & McNamara. In private practice, James and Larry have worked with many patient organizations to ensure their community's voices were heard by decision-makers. Relevant to our EL-PFDD meeting, they have been involved in helping plan and moderating three-fourths of the over 75 externally-led PFDD meetings.

Hear from them on:

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

Save the Date!!!

...for our 2024 Family conference!

June 29-30th 2024
Aurora, Colorado

HCU NETWORK AMERICA

- Free attendance for families!
- Low protein & regular menus provided!
- Kids & Teens programs!
- Fantastic opportunity to meet other families living with HCU!
- Informative sessions & workshops!
- Learn about the latest in research for HCU!
- Vendor freebies!

more info coming soon!

HCU HEROES

RACE FOR RESEARCH

WALK / RUN / RIDE

HEROES IN THE MAKING

SEPTEMBER 1-30, 2023

HCU NETWORK AMERICA

aeglea

BE A HERO, RACE FOR RESEARCH

Cystadane; the last FDA approved drug for homocystinuria was approved in 1996. That was 27 years ago!

Currently, there are two 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 5 research grants, with two pending! 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!

SWAG INCLUDES

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

HCU HEROES

RACE FOR RESEARCH

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HEROES IN THE MAKING

SEPTEMBER 1-30, 2023

HCU NETWORK AMERICA

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8 Tips for a Successful Virtual Race (Tips 5–8)

#5 – Control what you can

#6 – Take charge of your timing

#7 – Be mindful of trail etiquitte

#8 – Raise funds & share your experience

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

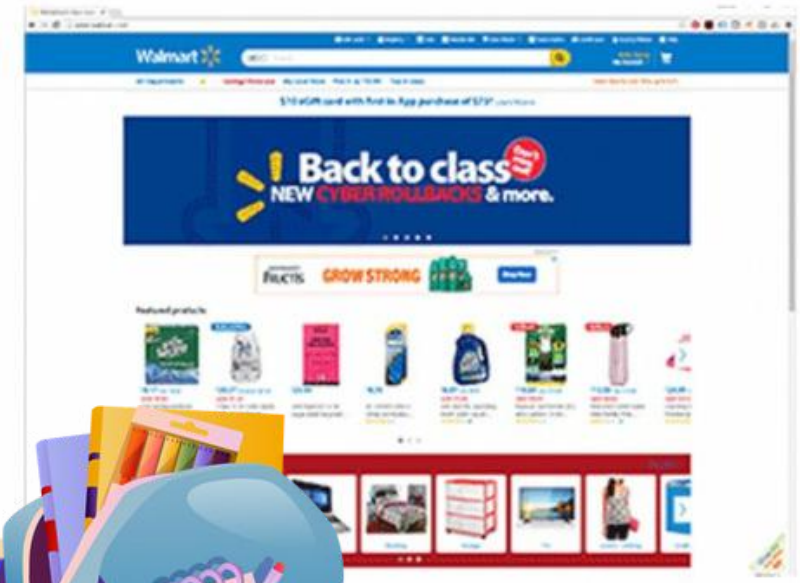
iGive.com

You Shop. Your Charity Gets Money. For Free.

Your Back to School Shopping makes a difference!

Shop using the iGive, naming HCU Network America as your charity, and iGive will donate to HCU Network America!

Want to make your purchases count?



The Button, iPhone, iPad, & Android apps make helping simple. Mouse over the image to see.

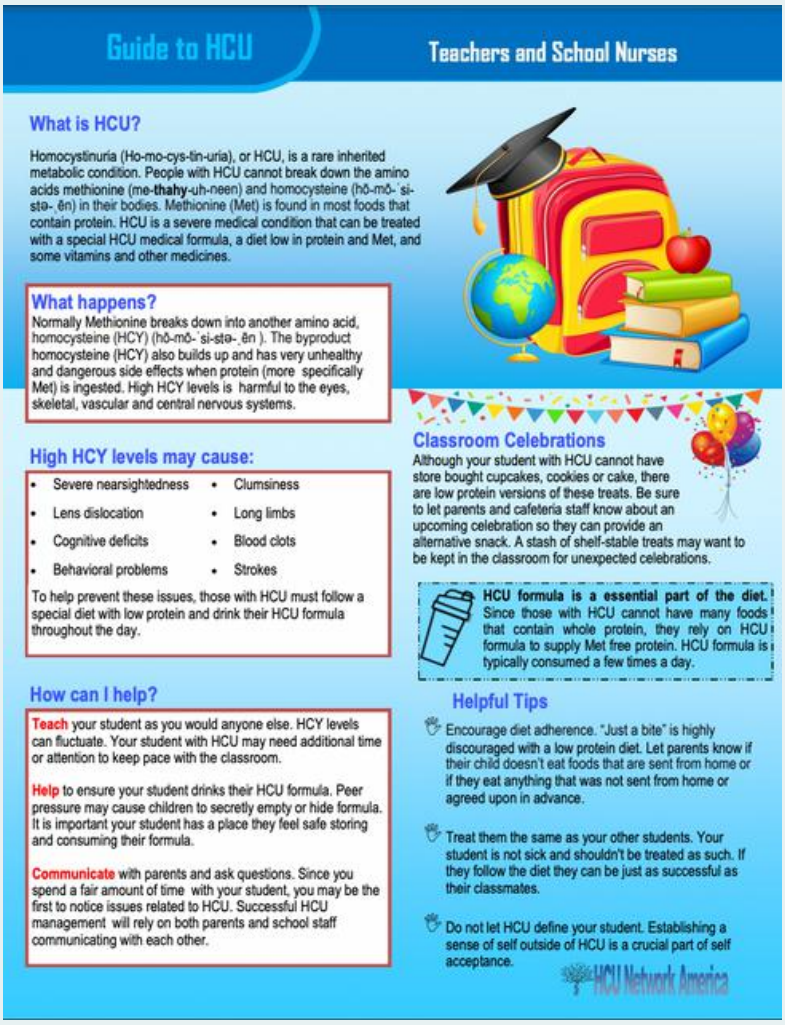
The optional iGive Button is a simple web browser app, easy to install and uninstall. It automatically activates at participating stores.

Don't want the Button or an app? Just start your shopping trips by going to iGive.com.

Shop normally (no special codes, no special anything) at any of about 1,900 stores. The Button is working in the background to let them know you're helping when you shop.

Head to iGive.com to sign up & start shopping!

We have Back to School resources!



Guide to HCU

Teachers and School Nurses

What is HCU?

Homocystinuria (Ho-mo-cys-tin-uria), or HCU, is a rare inherited metabolic condition. People with HCU cannot break down the amino acids methionine (me-**thahy**-uh-neen) and homocysteine (hō-mō-ˈsi-stə-ēn) in their bodies. Methionine (Met) is found in most foods that contain protein. HCU is a severe medical condition that can be treated with a special HCU medical formula, a diet low in protein and Met, and some vitamins and other medicines.

What happens?

Normally Methionine breaks down into another amino acid, homocysteine (HCY) (hō-mō-ˈsi-stə-ˌēn). The byproduct homocysteine (HCY) also builds up and has very unhealthy and dangerous side effects when protein (more specifically Met) is ingested. High HCY levels is harmful to the eyes, skeletal, vascular and central nervous systems.

High HCY levels may cause:

- Severe nearsightedness
- Lens dislocation
- Cognitive deficits
- Behavioral problems
- Clumsiness
- Long limbs
- Blood clots
- Strokes

To help prevent these issues, those with HCU must follow a special diet with low protein and drink their HCU formula throughout the day.

How can I help?

Teach your student as you would anyone else. HCY levels can fluctuate. Your student with HCU may need additional time or attention to keep pace with the classroom.

Help to ensure your student drinks their HCU formula. Peer pressure may cause children to secretly empty or hide formula. It is important your student has a place they feel safe storing and consuming their formula.

Communicate with parents and ask questions. Since you spend a fair amount of time with your student, you may be the first to notice issues related to HCU. Successful HCU management will rely on both parents and school staff communicating with each other.

Classroom Celebrations

Although your student with HCU cannot have store bought cupcakes, cookies or cake, there are low protein versions of these treats. Be sure to let parents and cafeteria staff know about an upcoming celebration so they can provide an alternative snack. A stash of shelf-stable treats may want to be kept in the classroom for unexpected celebrations.

HCU formula is a essential part of the diet. Since those with HCU cannot have many foods that contain whole protein, they rely on HCU formula to supply Met free protein. HCU formula is typically consumed a few times a day.

Helpful Tips

- Encourage diet adherence. "Just a bite" is highly discouraged with a low protein diet. Let parents know if their child doesn't eat foods that are sent from home or if they eat anything that was not sent from home or agreed upon in advance.
- Treat them the same as your other students. Your student is not sick and shouldn't be treated as such. If they follow the diet they can be just as successful as their classmates.
- Do not let HCU define your student. Establishing a sense of self outside of HCU is a crucial part of self acceptance.

HCU Network America

- **Educators Guides**
  Haz clic aquí para descargar en español
- **Navigating Accomodations**
- **Low Protein Lunch resources**
- **College Guide**

REGISTERING FOR ACCOMODATIONS

Many students with HCU don't register with Disabilities/ Auxiliary Services office at their school – but why not?! Some students simply don't know that it's an option and others are just completely turned off by the term "disability". We hear you, but give us a few minutes to explain why we suggest you register.

Reasons to Register:

1. Plan Before and Emergency
If you aren't registered with Disability Services, you aren't guaranteed any sort of accommodations for problems that may arise due to your HCU. You cannot register for accommodations retroactively. If you fail/miss a test because of a medical situation and you haven't registered, you'll have to accept the grade.

2. Don't Sell Yourself Short
It is scientifically proven that high homocysteine levels can seriously affect your academic performance. Perhaps you can get a passing grade on a test while your homocysteine level is high but imagine what you can get if it wasn't. Set yourself up for success.

3. Advocate for Yourself
Most professors are extremely accommodating, but once in a while you will encounter a professor who isn't. Registering with Disabilities Services overrides whatever rules your professor has established around food and drinks in the classroom, preferential seating, copies of power points, the ability to make up exams, etc.

4. Don't Let HCU Limit You
Accommodations can also extend to housing, dining, and registering for classes. This can include where you live, roommate situations, meal plans, and early class registration.

How to Register

1. Find out is in Charge of Accomodations
Each campus is different. The office in charge of accommodations could be called Disabilities Services, Auxiliary Services, Accessibility Services, or something similar. Try searching "Disabilities Services + (Your campus name)" to find the office you should connect with.

Still unsure? Try checking in with the health center, residential life staff, or student affairs.

2. Register Before You are on Campus
Reach out to the office responsible for accommodations as soon as you know which school you will be attending. Many campuses have a deadline for any special accommodation requests., so don't miss it.

3. Provide Appropriate Documentation
While every campus is different, it's a good idea to have the following documentation from your doctor:

- A diagnosis of your HCU along with it's symptoms
- An explanation of how your HCU is a disability

Click here to check out our Back to School & special edition topics!

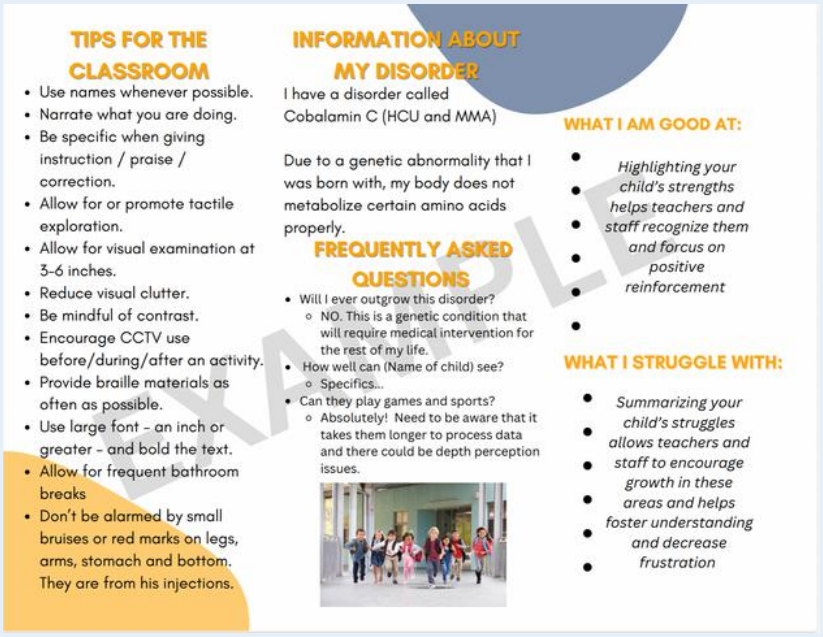
We have Back to School resources!

New Resource alert!

Customizable "All About Me" pamphlet

Designed for those with Cobalamin Disorders or Severe MTHFR

a great tool for teachers/school staff, coaches, babysitters & family members!



Click here to download & get started customizing your pamphlet!

New Resource

Let's help you *Transition to Adulthood*!

We're adding more resources to supplement our *Transition to Adulthood* milestones guide!

Medical Appointments

I can meet & communicate independently with my medical team during visits.

CLICK HERE

to download this resource

Click **here** to access our *Transition to Adulthood* guide & additional resources!

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: Zucchini Bread & Banana Slices
Lunch: Grilled Cheese & Tomato Soup
Dinner: Mediterranean Vegetable Casserole

T

Breakfast: Cinnamon Buns & Sliced Peach
Lunch: Taco Salad & Diced Watermelon
Dinner: Make Your Own Pizza

W

Breakfast: Yogurt w/Granola & Blueberry
Lunch: Avocado Toast w/Citrus & Mint
Dinner: Drunken Noodles

T

Breakfast: Crepe w/Nutella, Strawberries & Blueberries
Lunch: HotDogz w/Cucumber Slices & Grapes
Dinner: Jackfruit Kabobs w/Rice & Greenbeans

F

Breakfast: Bagel & Cream Cheese w/Strawberry Slices
Lunch: Roasted Ancho Veggie Tacos
Dinner: Asian Stirfry

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.

Mexican Corn Salad

Makes 3.5 servings | Serving Size 4 oz | 1.5 g protein per serving

Ingredients:

- 4 oz. Penne, Cassava
- 1/2 c Fire Roasted, Corn, Peppers and Onions
- 2 TBSP Scallions, green parts, thinly sliced
- 1 TBSP Cilantro, chopped
- 28 g Follow Your Heart Parmesan Style Shredded

Dressing:

- 2 TBSP Boursin Dairy Free Spread
- 1/2 tsp Chili Powder
- 1/4 tsp Ground Cumin
- 1 tsp Lime Zest
- 1 TBSP Lime Juice
- 1/4 c Mayonnaise

Directions:

1. Cook penne as directed on the package. Drain and rinse with cold water to stop the cooking. Set aside in a medium to large bowl.

2. While the pasta cooks, combine all ingredients for the dressing in a glass bowl. Whisk well to combine. Refrigerate until ready to use. This will allow flavors to blend and settle.

3. Once ready, combine all ingredients into the bowl with the penne. Gently toss to combine. Can be served right away, but the flavors can come together if it is refrigerated for at least 30 minutes.

Notes:
This can be prepared a day or two ahead of time (Although best to cook the pasta the day of serving so that texture is desirable). You can also leave out the pasta and add the rest of the ingredients to lettuce with a few tortilla chips for a simple salad.

UPDATE

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

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

What is the purpose of this program?

Having a rare disease is difficult. Adding in the complex care required to treat or manage that disease and figuring out how to pay for it makes a rare diagnosis even harder.

NORD's Classical HCU Medical Assistance Program offers eligible individuals diagnosed with Classical Homocystinuria financial support to pay for the low protein foods and physician prescribed medical formulas necessary in managing this HCU diagnosis.

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

MEDICAL ASSISTANCE

Who is eligible to apply?

This program is designed to help patients who:

- Have a diagnosis of Classical Homocystinuria.
- Are a United States citizen or U.S. resident of six (6) months or greater with evidence of residency such as a utility bill showing the patient's name and address.
- Meet the program's financial eligibility criteria.

What is the application process?

Patients may be referred to the program by their health care provider, their case managers, or they may self-refer.

A NORD Patient Services Representative will guide the applicant through the application process and verify eligibility for inclusion in the HCU Program.

Awards are based on meeting eligibility criteria, funding availability, and are made on a first-come, first serve basis.

NORD is Here for You

NORD, a 501(c)(3) organization, is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.

NORD was founded by families struggling to obtain access to treatments and whose advocacy for change led to the passage of the Orphan Drug Act in 1983. NORD assists eligible patients (those with medical and financial needs) in affording the treatments and medical services their healthcare professionals have prescribed.

Alone we are rare. Together we are strong.®

How do I get more information and apply?

Contact NORD's Classical HCU Medical Assistance Program

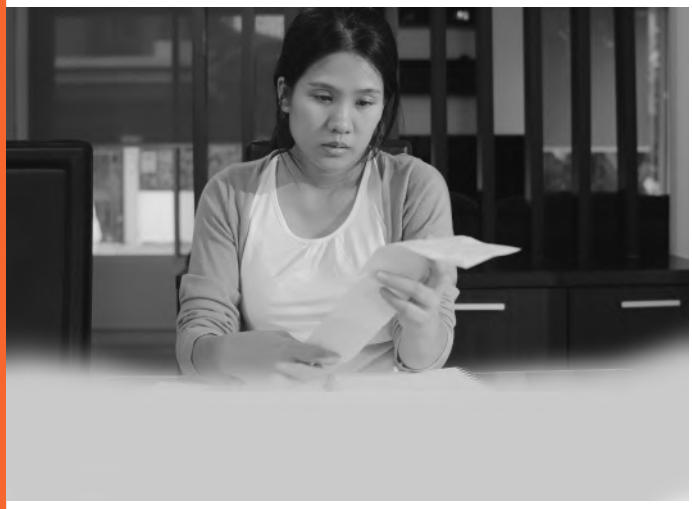
Monday-Thursday 8:30am – 7:00pm ET
Friday 8:30 am – 6:00pm ET

203-616-4327

203-635-4163

hcu@rarediseases.org

US MAIL to: NORD
Attention: HCU Program
55 Kenosia Avenue
Danbury, CT 06810



What assistance does NORD provide?

- The Classical HCU Medical Assistance Program assists eligible individuals with out-of-pocket costs to purchase low protein foods and physician prescribed medical formulas. Individuals approved for assistance in this program will be issued a PEX card. The PEX card is a prepaid expense card to be used for the purchase of low protein foods or physician prescribed medical formulas only.
- *Upon receipt of the card, the cardholder will contact NORD to request card activation. The card will be funded based on program award caps set for the program (this cap will be discussed with individuals upon enrollment in the program..*
  - *> It is necessary for the individual to submit receipts on a monthly basis evidencing card utilization for the purchase of low protein foods or medical formulas for the previous month.*
  - *> Funds will not be added to the card until the previous month's receipts have been received by NORD.*
  - *> The card may only be utilized for the purchase of low protein foods up to the monthly program limit.*

Once a patient is accepted into the assistance program(s) how long are they eligible?

Awards are issued for a calendar year.

Patients are encouraged to reapply annually if continued assistance is needed.

What happens if an applicant does not meet the criteria of the Electronic Income Verification?

The NORD Patient Services Representative will offer to e-mail, fax, or mail the brief program application and disclosure forms to the patient. The applicant may then complete the application, sign the disclosure form, provide the appropriate financial documentation to verify financial need, and return them via fax, email, or USPS mail.

How does NORD demonstrate compliance with regulations required of charities?

- NORD independently designs its patient assistance programs based on the needs of specific patient communities.
- No pharmaceutical company or donor controls or influences our programs.
- Our patient assistance decisions are based on consistently applied financial eligibility criteria and diagnosis only.
- Patients have their choice of health care provider, treatment and treatment location, and can make changes at any time.
- Patients' privacy and well-being are priorities at NORD. We do not share or provide patient names or data with donors, nor do we disclose or identify donors to patients. Patients are able to make the choices that are best for them because NORD's assistance covers all FDA-approved products available for a diagnosis. Our programs also help with more than medication: patients can use their funds to pay for other physician prescribed services related to their diagnosis, such as laboratory and diagnostic testing, physical and occupational therapy, durable medical and adaptive equipment, and travel to medical appointments.

NORD®

rarediseases.org

Nexus Patient Services

Commited to making life a little easier for people with metabolic disorders through innovation, quality and customer service.

HCU EASY TABLETS

Easy To Measure ✓

11 HCU Easy Tablets equals 10g Protein Equivalent

Easy To Swallow ✓

Almond shaped tablets help for easier swallowing

Easy On The Go ✓

Convenient for formula consumption while traveling, at work, or at school

Request a Sample!

Now available through Edgepark

HCU Easy Tablets should be taken under strict medical supervision and are suitable from 3 years of age.

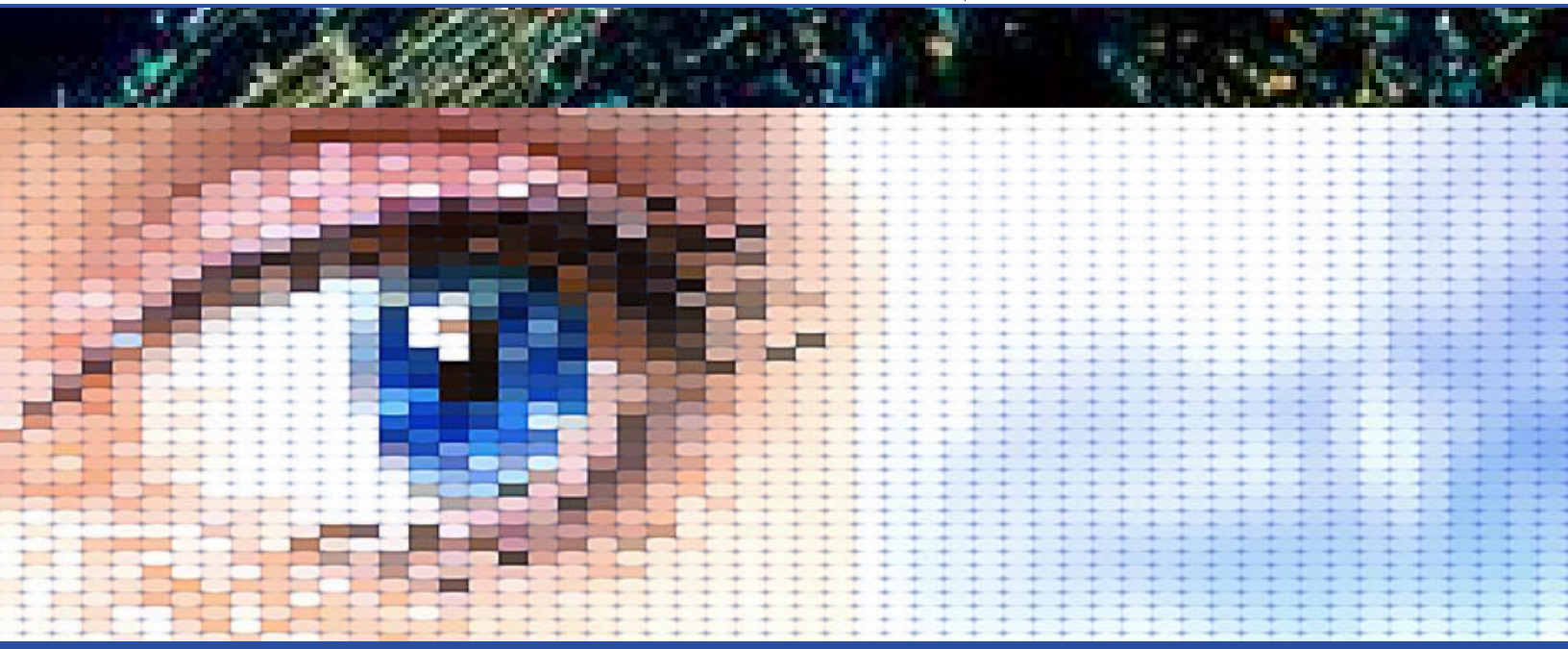
Nexus Patient Services now offers samples and patient assistance. Ask your dietitian for more information and a sample today.

Marketed and distributed by:

Nexus Patient Services
P.O. Box 15980, Phoenix, AZ 85060-5980
T: 833-875-0200
www.nexusmedicalnutrition.com

NPS-00031 JUNE 2022

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM



LET US SEE THINGS FROM YOUR PERSPECTIVE

From droopy eyelids (ptosis) to severe vision impairment, patients with various types of homocystinuria experience a wide range of eye issues. While some of these eye issues are better documented, other aspects aren't. We want to hear from you - help us see things from your perspective!

Complete the Eyes and Vision Survey

homocystinuria.rare-x.org

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE-X

HCU in the News / Industry News

Children's Hospital Colorado

Grayson: How Genetic Testing Saved His Life

It's rare to see the story of medical progress evolve within a single family. We don't always see how research, innovation and brilliant providers impact individual kids. When we do get a peek, it's literally lifesaving.

That's the case for Grayson and his family.

They felt the joy and relief that doctors and researchers can provide when they're willing to look through billions of pieces of genetic information. They also felt the profound sadness of having more medical questions than answers. Ultimately, their story and determination will help numerous children — some who aren't even born yet.

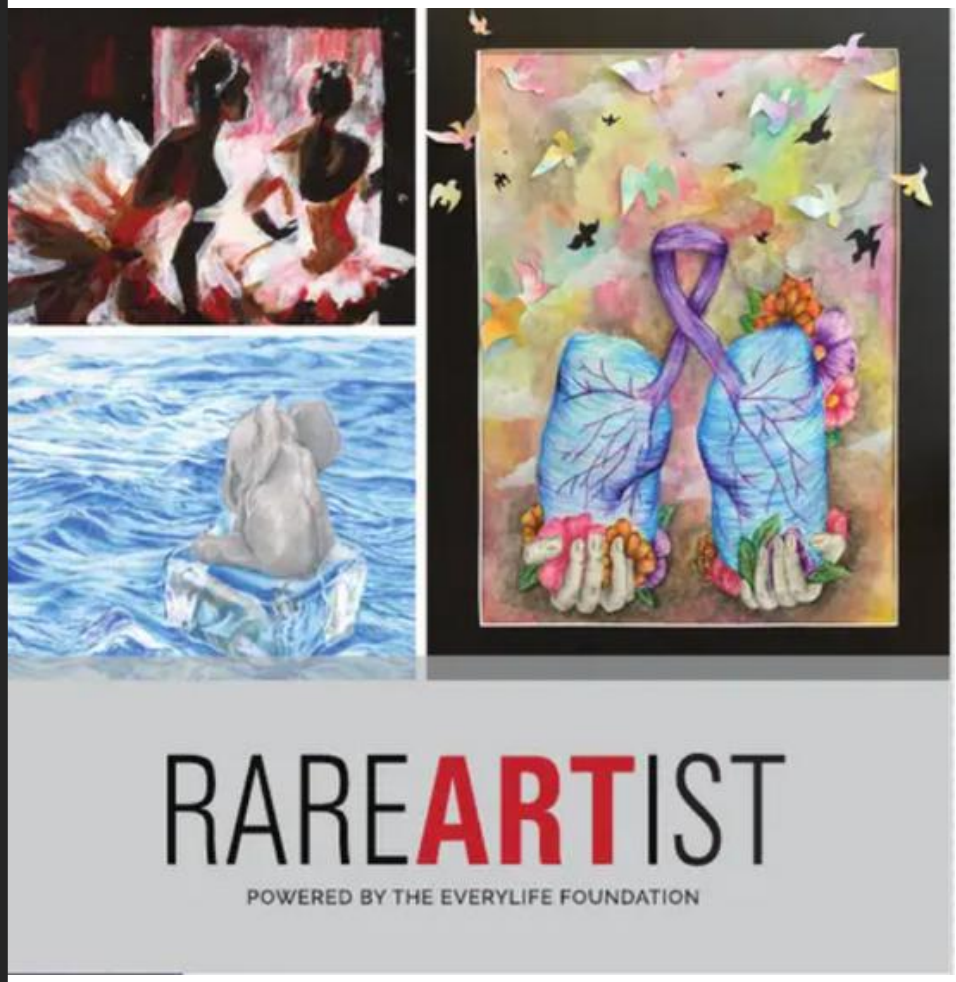
Click here to read the full story!

Codexis Announces Enhanced Strategic Focus and Extends Projected Cash Runway to Mid-2026

As a result of a strategic refocus announced by Codexis, they are discontinuing the development of an orally administered enzyme therapy for homocystinuria.

To read the full press release, visit https://bit.ly/3KcF99L

Calling all Rare Artists!



SHARE YOUR RARE THROUGH ART

Cash prizes awarded to winners.

Opportunity for art to be on display at Rare Disease Week on Capitol Hill.

Artist-to-Advocate individual coaching opportunity.

Contest is now open for submissions!

Click here for more information!

Submissions due August 31 at 5 pm

EVERYLIFE FOUNDATION FOR RARE DISEASES

Now Available!

Betaine Anhydrous for Oral Solution 180 gm

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

Copay support

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

Financial support

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

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

Eton Cares can provide copay and financial support.

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

Click Here

IMPORTANT SAFETY INFORMATION

Warnings and Precautions

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

Adverse Reactions

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

INDICATIONS AND USAGE

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

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

eTon PHARMACEUTICALS

Please see enclosed Full Prescribing Information for more information.

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

1402-v2

Acappella

NOW ENROLLING

ACAPPELLA Study on Classical Homocystinuria

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

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

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

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

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

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

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

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

If you have any questions, please email:

medinfo@travere.com

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

*Restrictions apply

MA-PE-22-0004. March 2023

TRAVERE THERAPEUTICS

Contact Register

What is the contact register?

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

What will this information be used for?

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

How do I participate?

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

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