The HCU Herald

Featuring...

RARE DISEASE DAY EDITION!

HCU Hero
Sarah from North Carolina

February 2023

HCU NETWORK AMERICA

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

HCU HERO: SARAH FROM NORTH CAROLINA

Introduction

Hi, my name is Sarah May. I am 16 years old and I live in Kernersville, North Carolina! I am a patient that is living with Classical Homocystinuria. I was diagnosed at birth through Newborn Screening, as a result of a high methionine level. I am here today to tell you about my life from beginning to up until now, and how I live an everyday life with such a unique and rare genetic condition. But before we dive in, I want to thank my Metabolic Geneticist and Nutritionist at the UNC Chapel Hill Genetics clinic located in Raleigh,NC and of course my family, who have been with me since birth, helping me to navigate through the hardships and find out who I really am.

The Beginning

Like I said above, I was diagnosed with Classical Homocystinuria through Newborn Screening. While I am sure that the news must have been scary for my parents, we now know what a huge blessing it was to get a diagnosis at birth. My parents had two children prior to me - my brother, Jackson (now 21) and my sister, Emily (now 19), so the news that I had a rare disorder came as a total surprise. My mom once told me this story which she now calls her 'testimony'. She said that she was on a trip with my dad and she kept having this sudden gut feeling that something was wrong with her pregnancy, so she went and checked in with her doctor when they were back in town. The doctors kept telling her that there was nothing wrong and that as far as they could tell, I was a super healthy baby. But my mom knew deep down that something just wasn't right.

HCU HERO: SARAH FROM NORTH CAROLINA

During the pregnancy, my parents experienced a move to North Carolina from where they originally lived in Abingdon, Virginia. To make a long story short, moving to North Carolina was what my mom thinks of as god's intervention, because if we had not made that move, my HCU would have not been detected through newborn screening due to the difference in newborn screening between NC and VA.

After I was born, I spent 6 weeks in the NICU and from there I was sent to the UNC Chapel Hill clinic to be under the care of a geneticist. My mom was immediately told to stop breastfeeding. She remembers saying to my doctor, "If you can just tell me how to feed her and tell me what I can pack her for lunch on the first day of kindergarten, I can figure this out." After that appointment, boy did my parents realize that they were in for a journey! Day by day my mom figured out new tactics and ways for me to take my medications. Next up was dealing with toddlerhood and then came the first day of big girl school! My mom felt that since I was young, she was able to watch over me and make sure that my food looked like everyone else's, made sure I fit in, and was eating the foods that were good for my diet.

But, as elementary school approached, my mom's anxiety grew. She worried about me feeling out of place or different than the other kids around me. But, as always, we got through this next phase of life as a family.

"...if we had not made that move, my HCU would have not been detected through newborn screening due to the difference in newborn screening between NC and VA."

HCU HERO: SARAH FROM NORTH CAROLINA

I am forever grateful for the love and support from my entire family, for example my grandma. As an infant my family (and baby me!) had to travel to Chapel Hill for appointments every three months, and as I got older that turned into every 6 months. My grandma was there for every step of the way - she never missed an appointment and was always there to be my mom's backbone.

Together, we entered into the next phase of life, which brought along with it many sports that intrigued me! I am now a teenager, and I think that this phase has made my mom the most anxious of them all. I say this because after going through this my whole life, I'm able to pick up on things. In this phase she has had to let me figure things out, which means learning to care for myself, and I know that has been very hard for her as a mother. It has been one long journey but we are here today and I am so beyond proud and grateful to say I am healthy and I wouldn't be the person I am without the help of my hardworking, fearless mom.

Living with HCU

As a small child, I never really struggled with negative feelings about my HCU. It wasn't until I entered elementary school and from there on that I began to notice it.

At this point, all I wanted was to be like the other kids in my class. No other kid had to take multiple medications, drink a formula (that doesn't smell great), or eat certain foods that weren't “real”. I always asked my mom what was wrong with me, because, of course, I didn't understand. I can remember having a conversation with my mom one morning where I told her, “Mom, I can't wait to go to heaven so I don't have to have this.”

As my childhood went on, it really worsened through the grades where I was able to better understand what was going on. I would sneak foods that weren't good for me just so I could be “normal”. My geneticist always told me that it was normal to have these feelings. It took a long time for me to really “get it”, but then it finally stuck somehow. My biggest struggle has always been and still is, taking my formula and betaine. They both have a really bitter taste and have a distinct smell. Other kids would ask me “What is that?” or say things like “Ew! That stinks.” I had to learn to accept the fact that HCU was a part of me, and this is the way life was going to be.

At about the age of 8 I started playing softball, and it has been a lifelong passion from then on! Softball was almost a freedom to me; it made me feel normal. But I always had to consider HCU along with playing sports whether it be taking my formula before a game, or being careful with my bones because of being perhaps more susceptible to osteoporosis. As I finished middle school and entered high school, I was confident! It has been a long ride and is not something I would say has been easy at all. Today, I’m a junior in high school. I know that as high school comes to an end, I will be experiencing a 'new normal' with HCU as I’m sure I’ll have changes in routine. But I am beyond ready to make these big life changes and excited for what the future brings!

My Successes

- I've been able to play the sport I love (softball) and in my last two high school seasons, I've played in the North Carolina Softball State Championships!
- I've been able to stay grounded and be confident within my own body and navigate through all life has thrown at me, including HCU of course!
- I have been recognized for being in the top 20 of my class for both freshman and sophomore year, and hoping to continue on!
- I made the volleyball team at my high school and have played all three years.
- Finally, I won't keep making a boring detailed list but... I have overall been able to establish a routine to keep myself healthy, which includes making sure that I am taking all of my medications.

While reading this list of my successes, I hope the readers (of all ages) are able to realize that they, too, can do this! Yes, there are going to be failures and hardships. But I have always learned to keep my head held high and keep pushing forward. To those of you struggling with the diagnosis of HCU and trying to live a 'normal' life with it, the future holds great things for you, too.

To all newly diagnosed HCU Patients

There are many words of wisdom and lots of advice that I could give to all patients having struggles with HCU. But here are just a couple that I can discuss in the little space and time I have here. I will start off with advice that I can give to adults parenting a child that has HCU: I encourage that you as a parent offer support and guidance for your child. After all, it is as new to them as it is to you! I also encourage you to try new things, make new foods, and find new recipes. The more options, the better! Also, reach out. There are plenty of parents and children who want to connect with others in the community, and this can make your journey a little easier.

Some advice I can give to children/teens and adults struggling with HCU is to be confident first of all. You are no different than anyone around you! Stay in a routine with your medication, make sure you are taking all of your formula and betaine, or whatever it may be to stay healthy. Lastly, don't let HCU define you! Find a hobby or sport that you love. For me, being active in sports that I love has helped to keep my mind from focusing on what makes me different.

Now I want to briefly talk about some things that have helped me to disguise the taste of my formula and betaine. I have tried every single type of formula there is and I find the one that works best for me is the HCU Express packets. I mix three of those with warm water at night and stick it in the fridge. The next morning, I mix in some Mio (water flavoring) which is strawberry-watermelon flavored and it tastes just like juice. With my betaine, I take seven scoops twice a day. I tend to mix it with ginger ale or anything fizzy to disguise the bitter taste and that has worked very well for me also! Parents, if your child is taking any pills and has a hard time swallowing them, my mom always mixed them in a spoon of applesauce or pudding. As a child, I never knew the difference. These are just a few suggestions that have worked great for me and might also work well for you!

Hope for the Future

I have a great feeling about the future for HCU and the patients and families involved. I have really enjoyed being able to experience going to conferences where I can meet other people my age (also younger and older!) and hear about their journeys. Hearing others' perspectives has helped me as a patient because it helps me feel at ease. I really encourage all patients and families, if possible, to join us in these conferences where you can experience new information and learn about new studies going on in the HCU world! These conferences are what have given me hope. I hope in the future that there could be a cure; whether it be a shot, gene therapy or any other option. Maybe not even a total cure, but something that makes taking my medication easier or takes away some of the bad taste from the betaine and the formula. I also hope that food distributors can think about the money spent on these special foods because it is not cheap at all compared to foods bought in grocery stores.

One last thing that I hope for this community is that we can identify ALL cases of HCU at birth, rather than some patients experiencing life threatening situations for them to figure out they have this condition later on in life. The future is bright for the HCU community, and it just continues to get brighter. Our community is full of strong people, and is a place where there is no judgment.

Stay Connected!

I encourage all families and patients to stay connected through the website or social media pages. This can help you stay in contact with other patients, hear about recent studies, discover new recipes and finally hear about events occurring within the community! It has been a pleasure being able to share my story personally with the community and I hope that after reading this I have been able to help you or lead you through guidance for your journey also! Always remember that we stand together and you are not alone. Thank you!

I'll leave you with some memorable moments with friends that I've made through HCU Network America!

~Sarah

HCU Network America
Organic Acidemica Association
Propionic Acidemia Foundation
2022 Conference
Bethesda, Maryland | June 25-26, 2022

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: Cookie Butter Banana & Blueberries
Lunch: Grilled Cheese & Tomato Soup
Dinner: Veggie Pot Pie & Salad

T

Breakfast: Egg & Cheese Sandwich & Grapes
Lunch: Greek Pasta Bowl & Pear
Dinner: Mushroom Fajitas

W

Breakfast: French Toast & Banana
Lunch: Avocado Tomato & Feta Sandwich
Dinner: Linguini w/Roasted Veggies

T

Breakfast: Nutella, Strawberry & Blueberry Crepe
Lunch: Veggie Nuggets & Pretezel Sticks
Dinner: Broccoli Cheddar Soup, Sandwich & Dessert

F

Breakfast: Bagel w/cream cheese & pear
Lunch: Veggie Noodle Soup w/crackers & sliced kiwi
Dinner: Make Your Own Pizza

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.

Jalapeño Rolls

Yields 10 - 12 rolls | Each roll is 0.8g protein

Ingredients:

- 68 g Fresh Jalapenos, about 3 peppers
- 3 strips Plant-Based Bacon
- 2 oz. Violife Just Like Cheddar Shreds
- 250 g Schar Puff Pastry Dough, one roll, taken out to warm up

Note:
You may be able to get up to 12 rolls out of the dough. If you don't want spicy, then use the mini sweet peppers in place of the jalapenos. You can omit peppers and make just bacon and cheese rolls. Can serve with sour cream or ranch. I used Morning Star bacon strips when I made this recipe. The *Hooray* brand bacon may cook a little differently.

Directions:

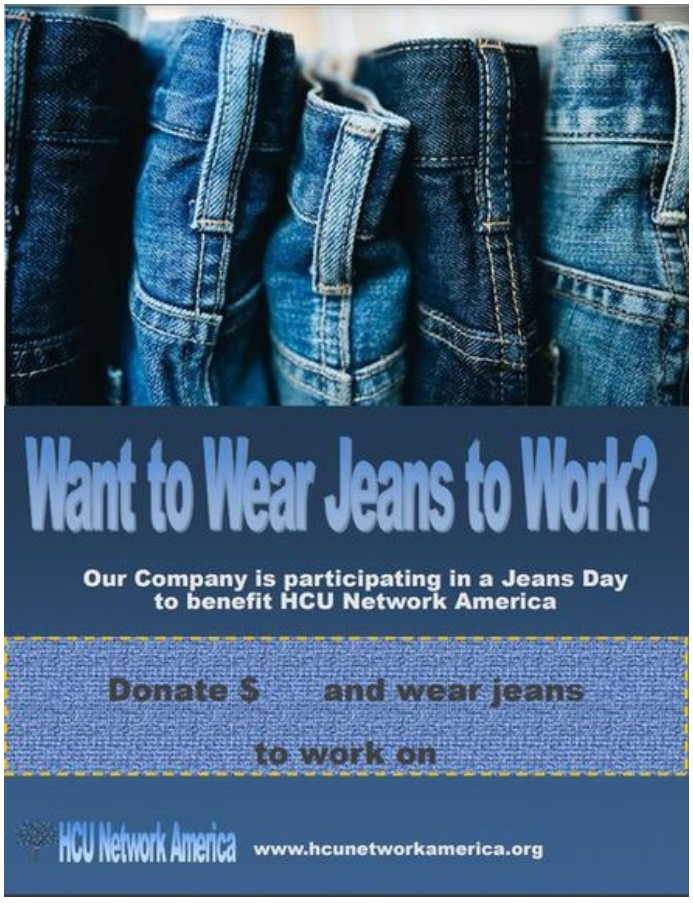
1. Prep oven or air fryer by preheating to 400 degrees. If baking in an oven, line a baking sheet with foil and spray w/cooking spray. Set aside.
2. Prep Jalapenos: Cut tops and discard. Cut the jalapenos in half lengthwise and remove seeds. Then cut each half in half again lengthwise to make four strips total per jalapeno. Prep bacon: Cut into strips the same length as the jalapenos.
3. Unroll the puff pastry. It can be delicate, so if it falls apart you can roll it into a ball and use a rolling pin to roll out again. Cut the puff pastry sheet into 3x4 inch strips. Does not have to be exact, just make sure the dough width is about same as length of jalapeno and bacon strips. Place one strip of jalapeno, one strip of bacon, and some nondairy cheese on the top of one pastry strip. Roll up and rub a little water at the end of the roll and pinch into the roll to seal. Repeat steps for all rolls.
4. **Air fryer:** Spray the rolls lightly with cooking spray and cook according to manufacturer's instructions. I fried mine at 400 degrees for 15 minutes, until nice golden brown in color and cheese was melted.
   Oven: Place rolls on prepared baking sheet and lightly spray with cooking spray. This will help with browning. Bake at 400 degrees for 12-15 mintes. Serve warm.

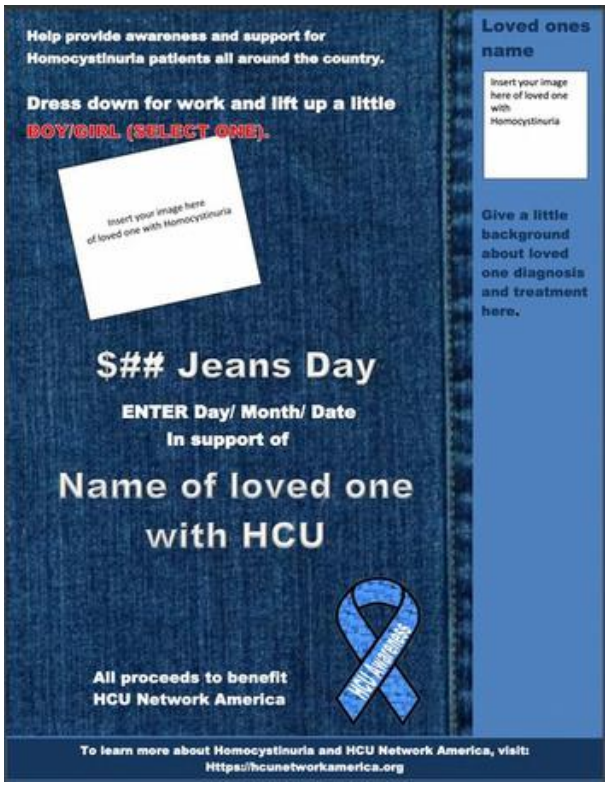
FEBRUARY FUNDRAISING EVENT: CASUAL FOR A CAUSE

Itching to forgo the traditional slacks and skirts for a more relaxed look at the office?

Take the lead in your company to sponsor a Casual Cause: Jeans Day to ***raise funding for the resources and tools HCU Network America*** provides to the Homocystinuria community!

Encourage employees to dress down for a day or even an entire week by requesting donations in exchange for a day in their casual best.





Not sure where to start? No problem! We've created several "Team Captain" materials to help you on your way toward sponsoring a successful fundraiser. Take a look at our customizable flyers, stock emails to inspire participation, tracking sheets and more!

View resources at:
https://hcunetworkamerica.org/casual-for-a-cause/

FEBRUARY FUNDRAISING

Grab your gear! https://www.bonfire.com/store/hcu-haberdashery/

Order by February 5 to receive in time for Rare Disease Day!

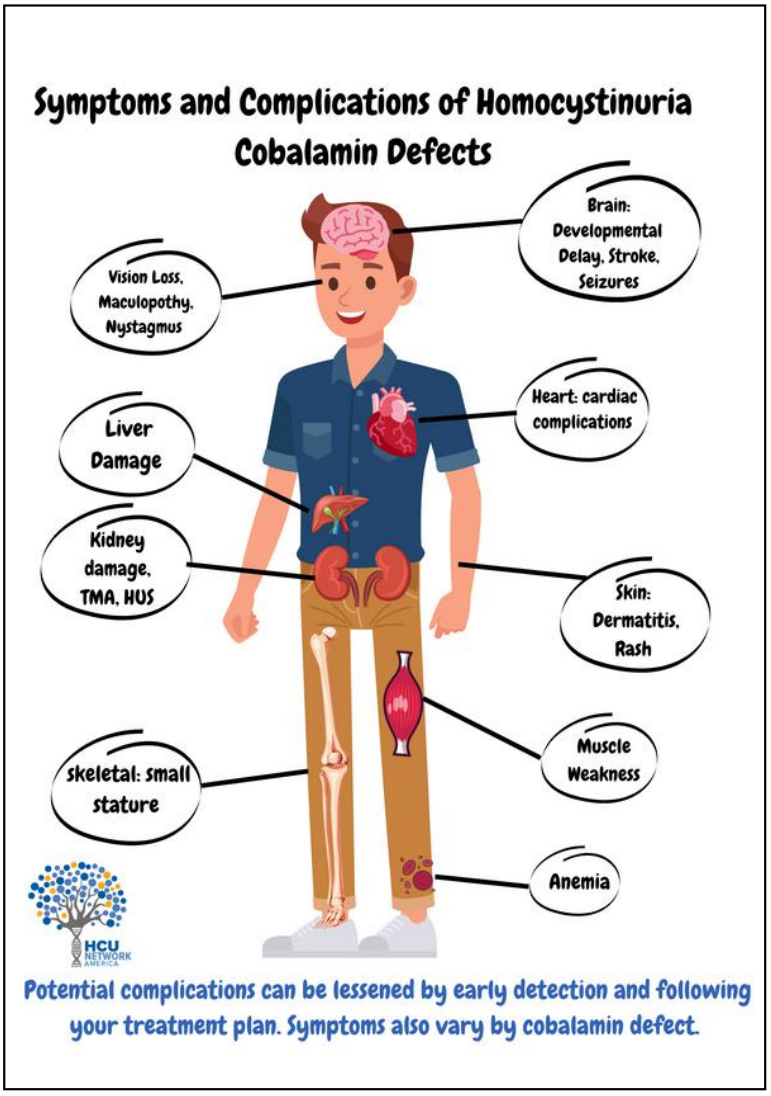
New Resources

Let's help you *Transition to Adulthood*!

In December we debuted our new Transition to Adulthood, Milestones Assessment Guide, and this month we are sharing some more resources to go along with it!

I understand the symptoms & complications of homocystinuria if left untreated.

These printable resources are designed to help you to better understand the potential manifestations of your specific kind of HCU, if left untreated.



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

Back to Care Program

Feeling like you need to hit the 'reset' button?

With all of the challenges that managing HCU presents, it's easy to stray away. But the good news is...

It's never too late to get back on track.

Click to access our **Back to Care guide**

Click to learn about our **Back to Care mentor program**

> I knew that I needed to take care of ME so that I could have the future that I didn't think I could have before.
>
> -Aimee from Maryland

RESEARCH

#HopeConnectsUs

It's more than just a hashtag - just look at this map!

It's the patients & caregivers from 26 US states & 14 countries who are connecting our community with hope by participating in our **Rare-X Data Collection Program**!

Be counted & Join the movement of hope!

Help us to Shine a Light on HCU by sharing your experiences.

REGISTER NOW!

https://homocystinuria.rare-x.org/

UPCOMING EVENTS

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

HCU NETWORK AMERICA

HOMOCYSTINURIAS DATA COLLECTION PROGRAM

RARE X

HEART AND BLOOD VESSELS FACEBOOK LIVE CHAT

FEBRUARY 22 AT 7 PM ET

Misty
Cobalamin C, Parent

Christa
Classical HCU Patient

Danae'
Classical HCU Patient

Samantha
Classical HCU Patient

HOMOCYSTINURIAS DATA COLLECTION PROGRAM

1. Enroll at (5 minutes)
2. Take the Health and Development Survey (15 minutes)
3. **Take the Heart and Blood Vessels Survey (10 minutes)**

Complete the Heart and Blood Vessels Survey at: homocystinuria.rare-x.org

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

UPCOMING EVENTS

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

Classical HCU Patient Virtual Meet-up

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

Sunday, February 5, 2023
2 pm CST | 3 pm EST

HCU NETWORK AMERICA

Classical HCU Parent-Caregiver Meetup

Parents,Grandparents and Caregivers of "kids" all ages with HCU need support too!

Saturday, March 4, 2023 | 10 am CT / 11 am ET

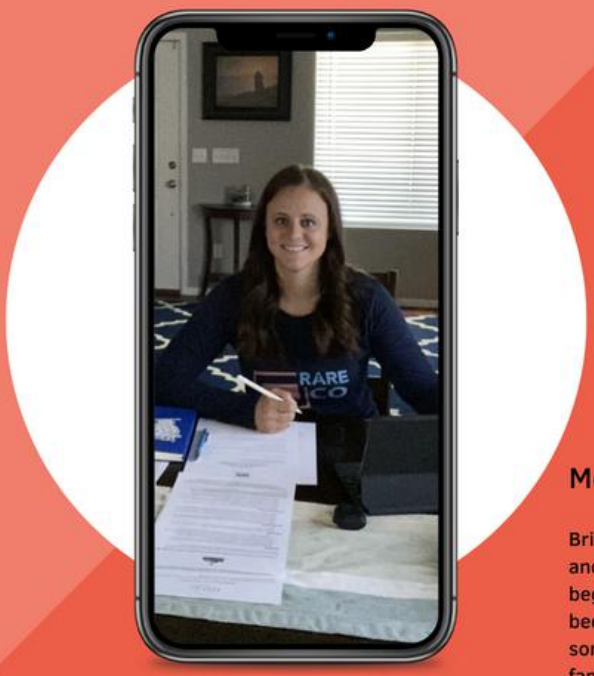
Meet your Meeting Facilitator!

Melanie is the mom of Masen (age 10) who was diagnosed with Classical Homocystinuria last year on June 25, 2021. Masen had to make big changes in his life at the age of 8 years old after this very surprising diagnosis. Masen underwent two eye surgeries (one on each eye) this past April where they removed the lenses from his eyes due to the damage that had been done with so many years being undiagnosed. He now wears contact lenses daily and is seeing great! Masen and family live in Vancouver, BC in Canada.

HCU NETWORK AMERICA

UPCOMING EVENTS

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



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.

Saturday, February 25 2023
12 noon EST

Meet your meeting facilitator:

Brittany Parke lives in the Denver suburbs of Colorado with her husband, Robert and three children, Alexis, Riley, and Grayson. While their family is just beginning the journey with Grayson who was diagnosed with Cbl G, they have been involved in the rare diseases community since the birth and death of their son Drew in 2011. Brittany loves to read, run and spend time outside with her family.






COBALAMIN STEERING COMMITTEE

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

JOIN US!

Help drive the future of COBALAMIN DISORDERS

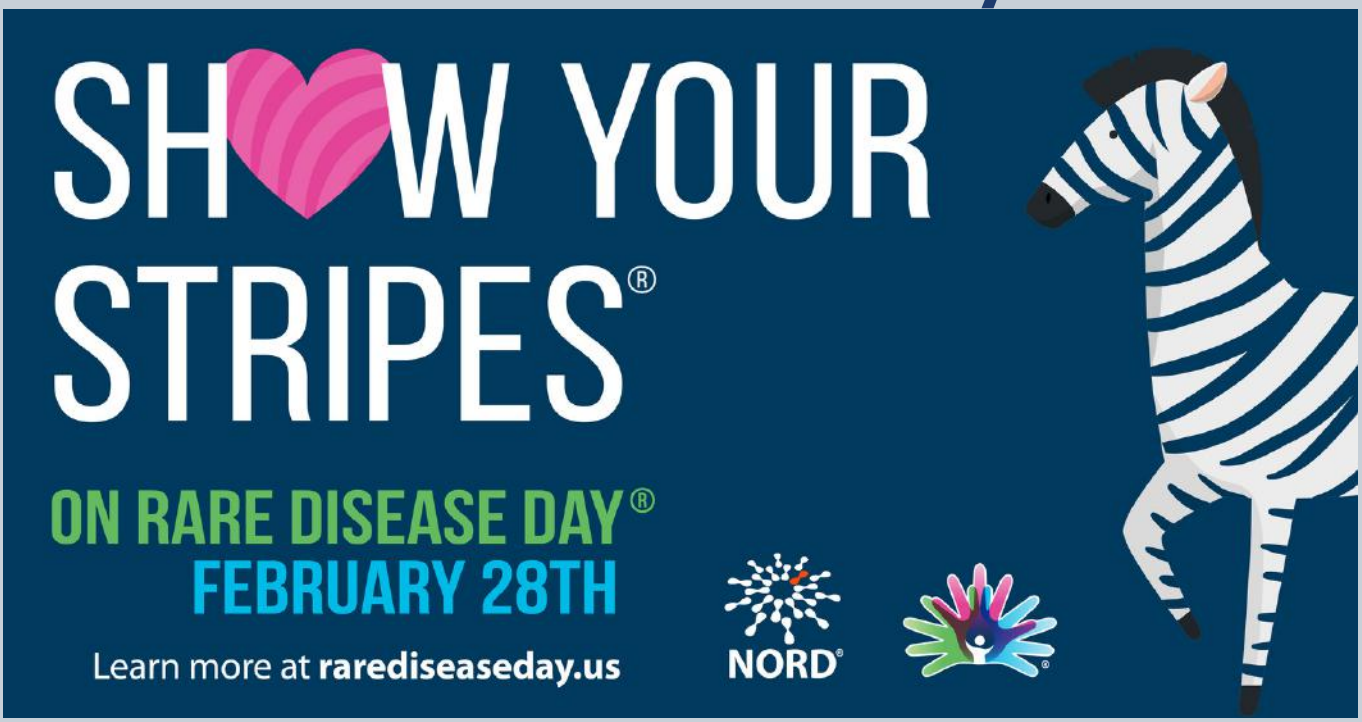
LEARN MORE ABOUT THE COMMITTEE AT:
http://bit.ly/HCUNACblSC OR
EMAIL: cblsc@hcunetworkamerica.org

HCU NETWORK AMERICA

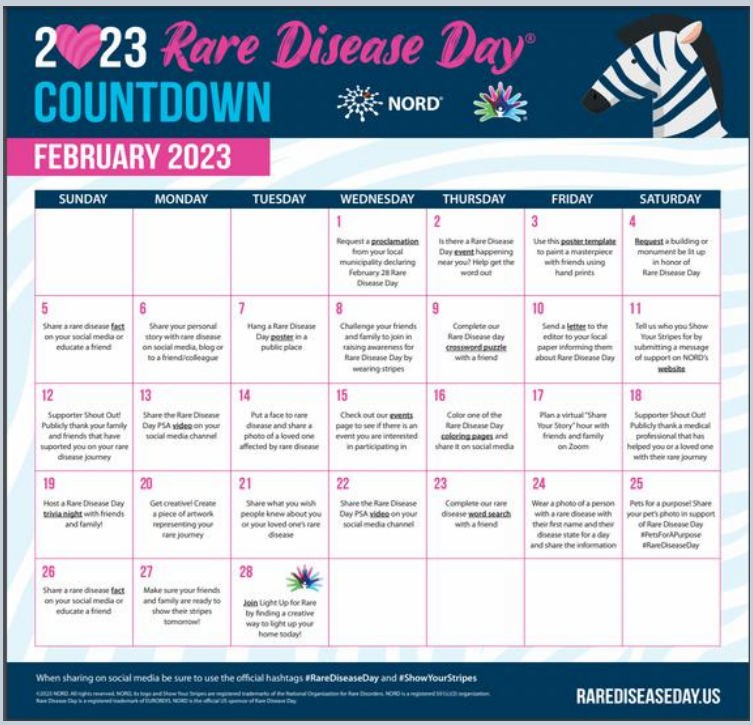
UPCOMING EVENT

MARK YOUR CALENDARS...

...for Rare Disease Day 2023!



Want to get involved but not sure where to start?



Click here to download this fun Rare Disease Day activity calendar!

UPCOMING EVENT

Now in its 12th year, **Rare Disease Week on Capitol Hill** empowers and inspires hundreds of advocates each year. The connections you make during the week will impact rare disease patients for generations to come.

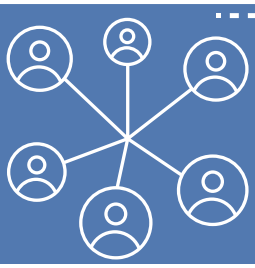
Hosted by the Rare Disease Legislative Advocates (a program of the EveryLife Foundation for Rare Diseases), the multi-day event brings together rare disease advocates from across the country to make their voices heard by their Members of Congress. Participants are educated on policy proposals impacting the rare disease community and provided opportunities to advocate for policy changes directly to their Members of Congress.
No matter one's connection to rare disease or their advocacy experience level, all are welcome.

To learn more, and to register to attend visit:
https://everylifefoundation.org/rare-advocates/rare-disease-week/

LET US KNOW!

Planning to attend Rare Disease Day events?

We'd love to connect you with others in our community who will be attending!



UPCOMING EVENT

NORD | RARE. STRONG. TOGETHER. 40 YEARS

Alone we are rare. Together we are strong.®

ABOUT | NEWS | EVENTS | CONTACT | DONATE

Will you join us for *NORD's 2023 Living Rare, Living Stronger Patient and Family Forum* on **Saturday, May 6,** 2023 in Washington, DC?

This patient and family-focused event provides an educational, immersive, and connection-driven experience for all attendees. Hear real stories from our community and gain practical knowledge to help you and your loved ones live your best rare life.

Topics likely to include:

- Finding and Living Your New Normal with a Rare Disease
- Drug Development for Rare Diseases
- Rare Breakthroughs: Now and On the Horizon

We are proud to offer **free registration** for patients and caregivers! This event includes an opening reception, a full day of programming, breakfast, lunch, event swag, and more.

For patient and caregivers needing travel & lodging assistance to attend, please review the guidelines and apply here.

**Registration should not be completed until you are notified of acceptance.*

UPCOMING EVENT

Midwest Genetics Network
Region 4

Register Now!

TRAUMA-INFORMED CARE IN THE MEDICAL GENETICS CLINIC

FREE WEBINAR:

February 23, 2023
12:00 CT/1:00 ET

CE Credits Available

OBJECTIVES

- Understand how trauma impacts children & families with genetic conditions
- Explain ways to recognize trauma
- Develop skills to support those with or at-risk for trauma and trauma responses

SPEAKERS

Heidi Davis, APRN, CPNP
Pediatric Nurse Practitioner
Gillette Children's

Kelsey Sala-Hamrick, PhD
Clinical Psychologist
Michigan Public Health Institute

Jessica Scott Schwoerer, MD
Medical Geneticist
Medical College of Wisconsin

Click here to register for this free event!

CONFERENCE RECAP/REPORT

The HCU Network America 3rd patient-expert conference was held June 25-26, 2022, in Bethesda, MD. Over 150 patients, parents, relatives, medical professionals, researchers, and industry reps from 26 states and 3 additional countries came together to celebrate our Brave HCU Heroes!

Check out our page dedicated to the 2022 conference! You'll find the program, presentation summaries, videos, photo gallery, and MORE!

https://hcunetworkamerica.org/2022-conference/

Attention friends in Canada!

This message is for you.

Did you know that HCU is supported under the Canadian PKU and Allied Disorders umbrella?

HCU stands for Homocystinuria.

We are looking to meet people who have HCU and are living in Canada.

Tell us about the YOU in #HCYOU

Email HCU@CanPKU.org

#HCYOU

https://canpku.org/

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

MEDICAL ASSISTANCE

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 necessary in managing this HCU diagnosis.

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

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?

NORD's program can assist eligible individuals with the expense of purchasing low protein foods:

- The Classical HCU Medical Assistance Program assists eligible individuals with out-of-pocket costs to purchase low protein foods. 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 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 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.

Program assistance is dependent on funding availability.

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

INDUSTRY NEWS

Travere Therapeutics Provides Corporate Update and 2023 Outlook

In the Travere Therapeutics Corporate Update and 2023 Outlook, the company provided the update below on their pegtibatinase program.

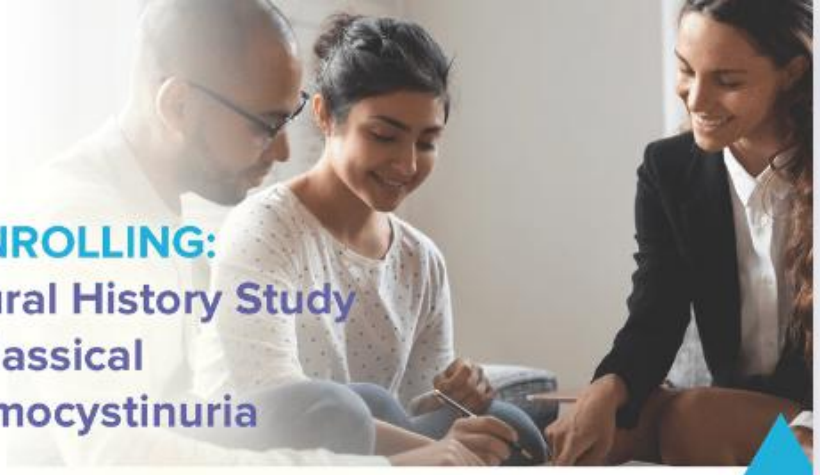
Pegtibatinase (TVT-058) – HCU
The Company continues to advance pegtibatinase, a novel investigational enzyme replacement therapy with the potential to become the first diseasemodifying therapy for people living with classical homocystinuria (HCU). Following positive results from the first five cohorts of the ongoing Phase 1/2 COMPOSE Study, the Company is evaluating pegtibatinase in a final cohort in the COMPOSE Study to further inform its potential pivotal development program. In the fourth quarter of 2022, enrollment completed in the sixth and final cohort of the ongoing Phase 1/2 COMPOSE Study. The Company anticipates reporting additional data from COMPOSE in mid-2023. In parallel with completing the final cohort in the COMPOSE Study, the Company is preparing for the initiation of a pivotal Phase 3 clinical trial of pegtibatinase in patients with HCU in the second half of 2023.

To read the full press release, visit: https://bit.ly/3Zt6PwV

Synlogic Provides Corporate Update and Outlook for 2023

Synlogic Therapeutics announced that in addition to a Fast Track designation and Orphan Drug Designation (ODD), SYNB1353 for HCU was also granted the Rare Pediatric Disease Designation (RPDD). SYNB1353 is an engineered probiotic designed to consume methionine in the gut to prevent absorption and conversion to homocysteine.

To read the full press release, visit: http://bit.ly/3HddfJu



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

DID YOU KNOW?

You could be eligible for a **paid clinical study** seeking to help future generations managing homocystinuria.
To learn more, visit: http://bit.ly/3WUgYQS

Participate in a Homocystinuria (HCU) Research Study

This study is looking at the safety of an investigational drug, pegtarviliase, and how well it is tolerated in patients with Homocystinuria (HCU)

About The Study

- Patients will participate across the globe
- Your participation will last approximately 14 weeks to include a screening period, 4 weeks of treatment, and a follow-up period
- You and your healthcare provider will know that you are taking the study drug (Open Label)
- Some study visits may be done at your home
- Study-related expenses (travel service, reimbursement for loss of earnings, and other study-related expenses) will be provided

Interested in Participating?

Please contact Kellyn Pollard and Juana Luevano at:

GeneticsResearch@utsouthwestern.edu

at The University of Texas Southwestern Medical Center in Dallas, TX

Why Participate?

- Help advance HCU research and therapies
- Help scientists understand how pegtarviliase works*
- You will be receiving pegtarviliase, a potential treatment before it is widely available

Who Can Participate?

- 18 years of age and older
- Diagnosis of HCU due to Cystathionine β-Synthase (CBS) deficiency
- For additional eligibility criteria, please scan the QR code below, or visit: https://bit.ly/3CX7XAd

*This study may be of no benefit to you. Taking part in this study may or may not improve your health.

aeglea.com

aeglea

Participe en un estudio de investigación sobre homocistinuria (HCU)

Este estudio analiza la seguridad de un fármaco en investigación, pegtarviliase, y qué tan bien se tolera en pacientes con homocistinuria (HCU)

Acerca del estudio

- Tendrá participación de pacientes de todo el mundo
- Su participación durará aproximadamente 14 semanas e incluirá un período de evaluación, 4 semanas de tratamiento y un período de seguimiento
- Usted y su proveedor de atención médica sabrán que está tomando el fármaco en investigación (Etiqueta abierta)
- Algunas visitas del estudio se pueden hacer en su casa
- Se cubrirán los gastos relacionados con el estudio (servicio de viaje, reembolso por pérdida de ingresos y otros gastos relacionados con el estudio)

*Puede ser que este estudio no sea de beneficio para usted. Participar en este estudio puede o no mejorar su salud.

¿Por qué participar?

- Para avanzar a ayudar la investigación y las terapias para HCU
- Ayudar a los científicos a comprender cómo funciona pegtarviliase*
- Recibirá pegtarviliase, un tratamiento potencial antes de que esté ampliamente disponible

¿Quién puede participar?

- 18 años de edad y mayor
- Diagnóstico de HCU por deficiencia de cistationina β-sintasa (CBS)
- Para conocer criterios de elegibilidad adicionales, escanee el código QR a continuación o visite: https://bit.ly/3CX7XAd

aeglea.com

aeglea

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

FOLLOW US