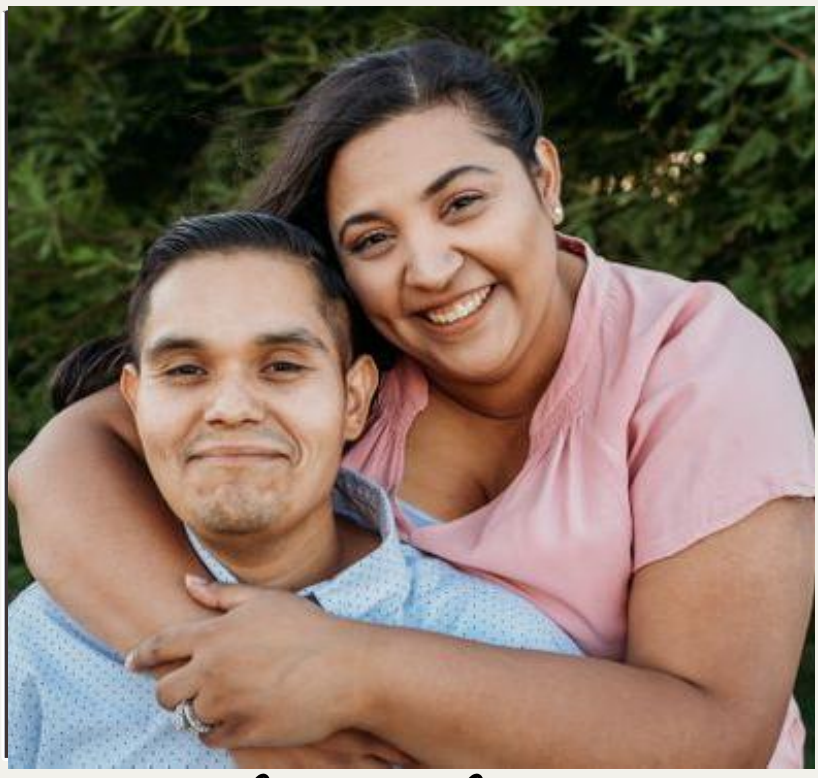
The HCU Herald

Featuring...



HCU Hero
Vicente from Arizona

April 2023

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

HCU HERO: VICENTE FROM ARIZONA

I'm Vicente and I was born in 1989 in Pheonix, Arizona. I was diagnosed with classical HCU through newborn screening at the Phoenix Children's Hospital. In fact, my twin brother and I were both diagnosed with HCU a few days after birth. Once we returned home, my parents got a call telling them that we needed to go back to the hospital for more testing. It was through this follow up test that they confirmed that my brother and I did indeed both have HCU. This is when my journey began.

Thankfully, my parents were able to provide us with formula and kept us on a strict low-protein diet. As we got older our family adjusted, always having something for us to eat at parties and family gatherings. They would make us feel welcomed and taken care of. However, starting school was a difficult part of my life. I would always take my lunch to school and never eat school lunch like the rest of the kids. It made me feel out of place, not to mention my low-protein food didn't taste the best, but I made it work. It helped that I grew up with the same friends from elementary school into high school, so everyone was understanding with me and didn't really question why I ate the way I did. They all knew about my disorder and I didn't really tell new friends about it until I got older. It seemed like the older I got, the more people were understanding and accepting of it.

HCU HERO: VICENTE FROM ARIZONA

As I finished high school and started working, I would only tell the coworkers that I talked to on a daily basis or went out to lunch with about my disorder. As I got older, I had a few relationships that didn't really go anywhere but in 2008 I met a woman that changed my life, my wife Stephanie. We met at work and started dating. I was really nervous to tell her about my disorder for some reason, so I kept quiet about it. I finally told her what was going on and she was accepting and understanding about the whole thing. Fast forward to 2012, we got married and today we have 3 kids. Thankfully, none of our kids were born with HCU and they are all in great health.

As an adult now living and working with HCU, everything just seems much easier than it was when I was growing up: from food options to formula and supplements. It's not as hard as it was growing up. I take my Betaine in the morning, make my Homactin shake, and carry my other 2 pouches in my lunch box with a snack. My wife has learned to cook a lot of low-protein meals and is always making something tasty for me to eat. Everything is falling into place in my life at this point and time, and I feel blessed to have been pretty successful in my career and life thus far.

"Everything is falling into place in my life at this point and time, and I feel blessed to have been pretty successful in my career and life thus far."

If you are new to the HCU community, whether you or your child has been diagnosed with this disorder, I would say to you: everything is going to be fine. There are tons of resources and alternatives out there in the world. Stay on diet and talk to your nutritionist and doctor. Use your resources so you are able to grow up and live a completely normal life. Take it a day at a time and everything will fall into place.

To read Vicente's story on our website, visit

EDIT THIS LINK

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

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

THE DISTRUBTORS OF HCU EASY TABLETS INVITE YOU TO

Enter to Win

TELL US ABOUT YOUR LIFESTYLE

*Click link above for a chance to win a $100 giftcard

Nexus Patient Services

833-875-0200
www.nexusmedicalnutrition.com

THIS WEEK'S MENU

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

M

Breakfast: French Toast w/Strawberries & Yogurt
Lunch: Veggie Nuggets & Celery/Carrot Sticks w/ranch
Dinner: Asian Stirfry Pasta & Garlic Bread

T

Breakfast: Bagels & Cream Cheese w/Sliced Fruit
Lunch: English Muffin Pizzas & Side Salad
Dinner: Tacos

W

Breakfast: Banana Muffin & Yogurt
Lunch: BLT Sandwiches & Chips
Dinner: Pasta Bake & Salad

T

Breakfast: Avocado Toast & Strawberries
Lunch: Burger & Pretzel Sticks
Dinner: Mac n' Cheese & Steamed Veggie Medley

F

Breakfast: Breakfast Sandwich, Banana & Blueberries
Lunch: Soup & Salad
Dinner: "Ricotta" lasagna, Garlic Bread & Side Salad

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.

Lemon Curd

Makes 2 servings | Serving 2 TBSP | 0.2g protein and 7 mg PHE

Ingredients:

- 40 g Lemon Pudding, dry mix, instant
- 1/2 c Sugar
- 1 TBSP Cornstarch
- 1/4 tsp Salt
- 1 c Lemon Juice
- 1/2 c Rice milk
- 1 c Canned Coconut Milk
- 1 TBSP Butter

Directions:

1. Combine dry ingredients in a medium saucepan. Whisk to combine. Turn heat to medium. Slowly pour lemon juice into dry ingredients while whisking to combine. Bring to a simmer and add the rice milk and coconut milk. Whisk and continue to cook until thickened. This may take a little time, but well worth it. The curd should be just thick enough to coat the back of a spoon. Remove from heat and stir in butter.

2. Pour the curd into glass jars or containers. Allow to cool to room temperature then refrigerate overnight. The curd may seem a little thin, but it will thicken once cooled and chilled overnight. Serve or use to your desire.

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®

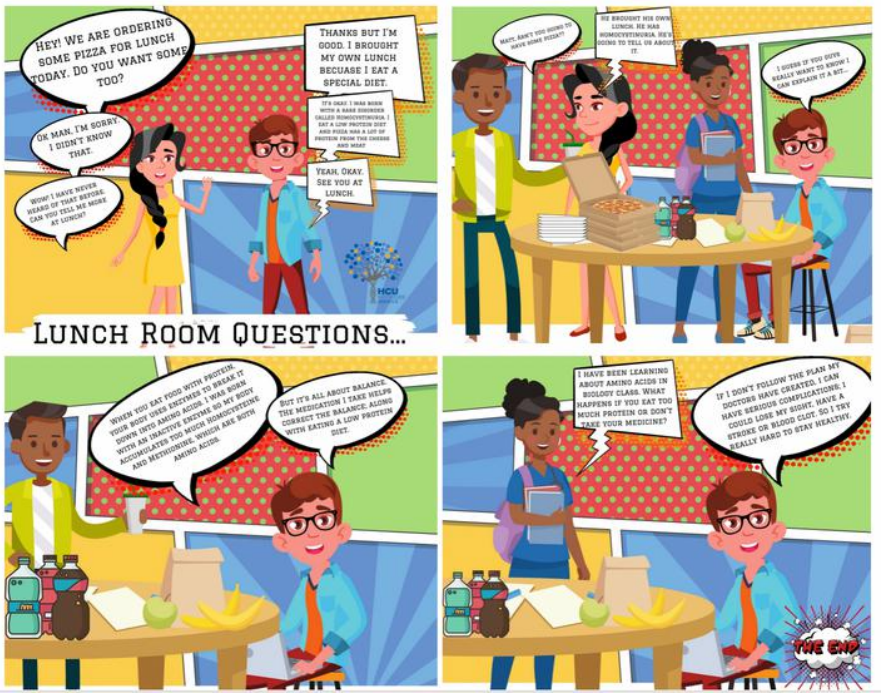
rarediseases.org

New Resources

Let's help you *Transition to Adulthood*!

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

I can explain the basics of my condition.



CLICK HERE

to download this resource for Classical HCU

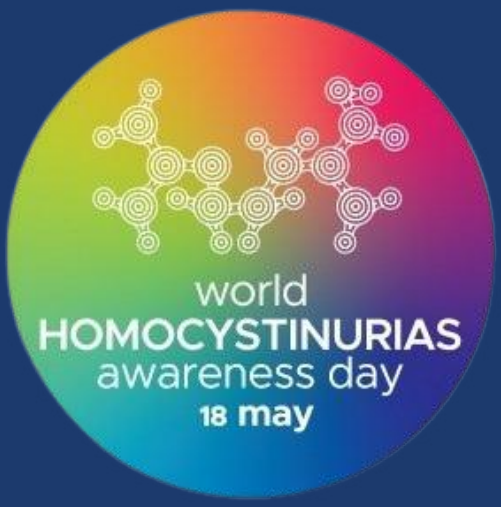
CLICK HERE

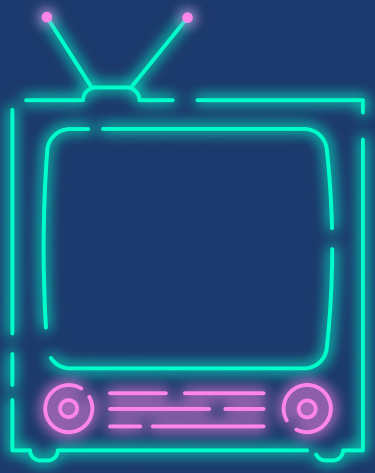
to download this resource for the Cobalamin Disorders

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

WE WANT YOU!:
WORLD HOMOCYSTINURIAS AWARENESS DAY

World Homocystinurias Awareness Day
is coming, and we'd like to feature YOU or your child in our promotional video!





Interested?

Email us @
mthfr@hcunetworkamerica.org

UPCOMING EVENTS

Find all events at: https://hcunetworkamerica.org/virtual-meet-ups/

Classical HCU Parent-Caregiver Meetup

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

Sunday, May 21, 2023 | 4 pm PT / 7 pm ET

HCU NETWORK AMERICA

Meet your meeting facilitator!

Danielle is a patient with Classical Homocystinuria diagnosed through the Newborn Screening Program in New York City. She resides in Winter Park, FL with her husband Irving, 3 sons and 3 dogs. Thankfully, having HCU is all she knows and has experienced minimal negatives and multitudes of positives. Danielle comes from a large family that serves as an amazing support system which continuously feeds her passion of servant leadership amongst the HCU community. Danielle is excited to lend her experience as a patient, traveler, foodie, and learning and development professional within the "big 5" health care companies to promote that living with a rare disease while simultaneously obtaining success in your passions is possible.

SEPTEMBER 1-30, 2023

HCU HEROES

RACE FOR RESEARCH

WALK / RUN / RIDE

More swag available for fundraisers!

Per Individual: $30

Per Family (up to 4 – 1 mailing address): $75

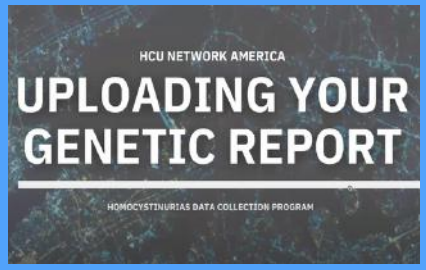
Save the date: Registration opens June 1, 2023

aeglea

HCU NETWORK AMERICA

RESEARCH

Our Rare-X Data Collection program continues to grow!



We need genetic reports!

Watch this video to learn how you can build a better understanding of homocystinuria by uploading your report!

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

HCU NETWORK AMERICA

HOMOCYSTINURIAS DATA COLLECTION PROGRAM

RARE X

SCHOLARSHIP OPPORTUNITY

Pursue Your Dreams through the #RAREis Scholarship Fund

Living with a rare disease means managing unique challenges, including frequent doctor visits, rigorous treatment regimens and hospitalizations, and exposure risks. While quality and duration of life continues to improve thanks to improved diagnosis and treatment approaches, individuals living with rare diseases still face disparities in achieving traditional life milestones. That's why the EveryLife Foundation for Rare Diseases established the #RAREis Scholarship Fund – to enrich the lives of adults living with rare diseases by providing support for their educational pursuits.

Thanks to the support of Horizon Therapeutics, The EveryLife Foundation for Rare Diseases will provide one-time awards of **$5,000 scholarships** to **35 rare disease recipients** in 2023.

Who can apply?

Anyone 17 or older, who is a resident of the United States and who has been diagnosed by a physician as having any form of rare disease, regardless of treatment status.

When are applications being accepted?

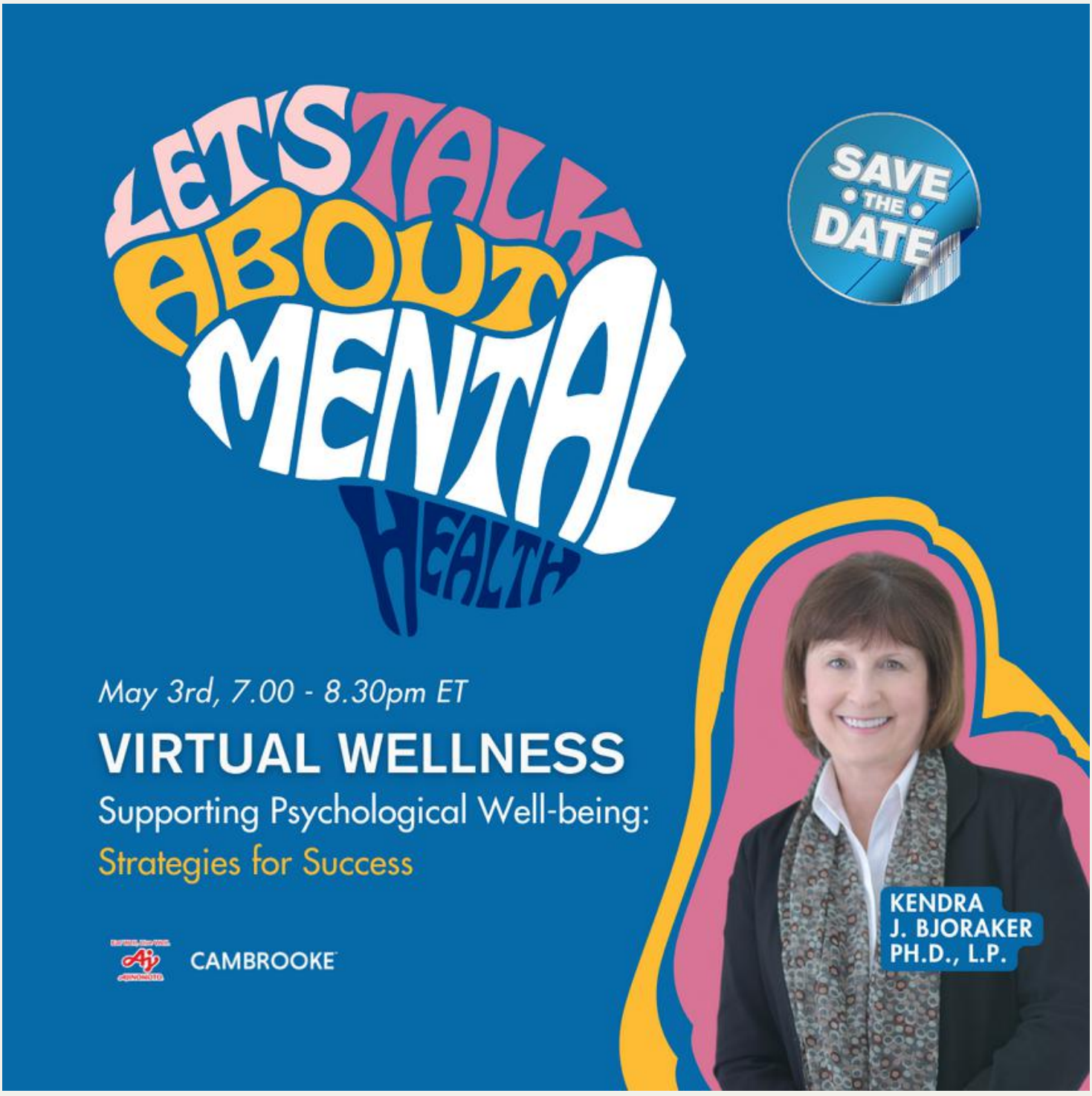
March 8- April 13, 2023.

What schools/universities qualify?

Applicants must plan to enroll full-time or part-time in undergraduate or graduate study at an accredited two- or four-year college, university, or vocational-technical/trade school for the Fall 2023 semester. There is no minimum amount of credit hours to be part-time. Students do not need to be pursuing an undergraduate or graduate degree.

Click here to learn more and to apply!

EVENT ANNOUNCEMENT



Click here to register for this event!

EVENT ANNOUNCEMENT

Family Camp, hosted by PKU News

WHEN?

June 1-4, 2023

WHERE?

Washington Family Ranch in Antelope Oregon

WHO?

for individuals with ANY inborn error of protein metabolism, their friends, family and caregivers.

Click here to find out more !

RECONNECT, BE A KID AGAIN!

A Kid Again.
Giving illness a time out

Where illness stops and Adventure begins!

A Kid Again provides Adventures for the entire family to give a break from hospital stays and surgeries

Learn more at www.akidagain.org

We make life for families caring for a child with a life-threatening condition“normal” again by helping them gain back moments of positive, family-shared experiences and memories.

A Kid-Again has two programs to offer, their Adventures program and ESports Team.

Learn more here:

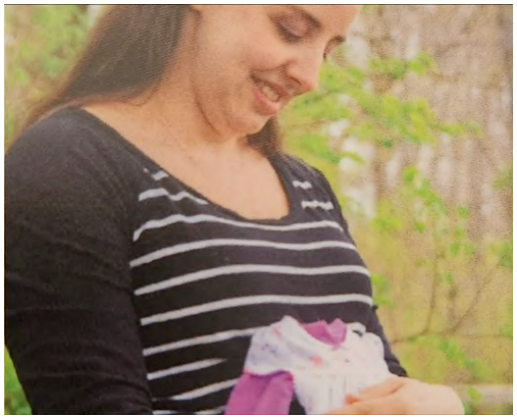
Adventure Program

E-Sports Program

HCU IN THE NEWS

Advocacy in Action:
Our community's own Amber Gibson was featured by her local new station!

21Investigates: Indiana tests for 35 of the recommended conditions in newborn screenings



FORT WAYNE, Ind. (WPTA) –In the state of Indiana newborns are screened for all but two of the conditions recommended for the screening panel. A mother who lives near Indianapolis tells 21Investigates she is thankful for that because her daughter's condition was caught weeks after she gave birth.

Many parents admit they do not know much about the conditions on the national Recommended Uniform Screening Panel (RUSP) that are aimed at ensuring equity in newborn screening from state to state. According to the Indiana Department of Health, there is an advisory board and standard process in place to evaluate the conditions that should be added for screening.

Indiana screens for 35 of the 37 conditions that are recommended. The only two that are not currently being screened have recently been added within the last year, according to a spokesperson from the state department of health.
Amber Gibson says she did not know much newborn screening, only that it was required by the state. She did not know exactly what was tested for. However, Gibson learned very quickly just how important newborn screening is.

"Had it not been for the newborn screen, we probably never would have known," she said.

Click here to read the full article & to watch the newscast video!

RESEARCH

BREAKING NEWS

Image courtesy of medpagetoday.com/

Researchers have developed a newborn screening test for HCU that measures homocysteine levels!

"Since 2006, the U.S. Department of Health and Human Services has included HCU on the list of disorders for which newborns should be screened. However, current tests only measure methionine levels, which are often still low when newborn screening occurs. As a result, these tests may miss an estimated 50% of HCU cases, which are then at high risk of being left untreated.

In order to remedy this situation, the researchers developed a newborn screening test for HCU that measures homocysteine levels. In infants with HCU, homocysteine levels usually rise before methionine levels. They almost always rise during the first few days of life -- the time when newborn screenings are usually performed -- making homocysteine a better early marker of this disease."

Read more about this exciting breakthrough!

http://bit.ly/3FE6Kye https://bit.ly/3JVvnsL

Your dollars at work: Updates from the 2022 CblC research grant!

Silvia Vilasi (Institute of Biophysics (IBF), National Research Council (CNR), Italy) Principal Investigator of the project "Identification of Compounds to Rescue MMACHC Functional Deficiency in CBLC Disease" granted by HCU Network America, Organic Acidemia Association and CblC Onlus Italian Association, tells us what objectives have been achieved during the first phases of the project and in which direction the research activities are going.

The long-term goal of our project is to find a novel pharmacological strategy to ameliorate CblC symptoms, exploiting a multidisciplinary approach that blends physics, chemistry and biology. To this end, we also need to better understand the impact of pathological mutations on the structure and function of MMACHC protein.
For these reasons during these first six months, we focused on the characterization of two pathological variants, selected together with Prof Carlo Dionisi Vici, Head of Clinical and Research Unit of Metabolic Diseases at the Ospedale Pediatrico Bambino Gesù, Rome (Italy) as representative of patients population. In parallel, we also started the screening of small molecules, with drug-like characteristics, able to bind MMACHC protein and hopefully revert the pathogenic phenotype.

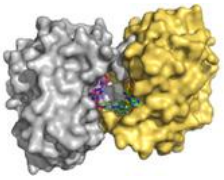


Figure 1: MMACHC protein dimers targeted with small molecules selected from a virtual library

By using molecular biology techniques, we paved the way for research by obtaining the isolated MMACHC protein, both in its functional form, the so-called wild type protein and two pathological variants resulting from the mutations identified in CblC patients. Then, we tried to answer some key questions: what is the structure of the mutated proteins, and how does it differ from the wild type, fully functional one? How do these proteins arrange themselves in the space? Are these mutant proteins able to maintain the correct shape when subjected to external stressors, i.e. are the mutated forms less stable then the wild type one? Can the mutated proteins still bind to another copy of itself (i.e. form a dimer) as observed for the functional protein? Are the mutated proteins able to bind vitamin B12, and promote its conversion?

We found that, compared to the wild type protein, the mutants weakly bind vitamin B12 and do not efficiently promote its conversion. In collaboration with Maria Grazia Ortore (Università Politecnica delle Marche, Italy) and Heinz Amenitsch (Graz University of Technology, Austria), we also observed structural issues: these labile mutants easily lose the correct conformation and do not form dimers. This "biophysical" characterization of the proteins is a fundamental step in order to understand which are the chemical-physical features, i.e., what kind of defect, therefore, should be corrected to ensure the restoration of the protein functionality. This is particularly true for the study of the protein's spatial three dimensional shape which is directly related to the protein function. All these data hint at a mutation-dependent destabilization of the protein architecture. Mutated MMACHC proteins adopt a suboptimal conformation which hinders its activity.

These structural defects can be countered by small molecules. Exploiting computational methods, we virtually tested 30'000 compounds and selected 11 predicted to bind MMACHC protein. The molecules were purchased and are currently being tested with the isolated proteins. Very preliminary data suggest that at least two of these candidates are able to bind the protein but we still need to understand if any of these molecules can restore functionality in protein mutants resulting from the CblC pathological mutations.

We are doing our best to provide at the end of the project indications on new molecules and/or new therapeutic strategies for CblC treatments".

Figure 2: Research team involved in the project at IBF CNR in Palermo

Figure 3: Research team involved in the project at IBF CNR in Milano

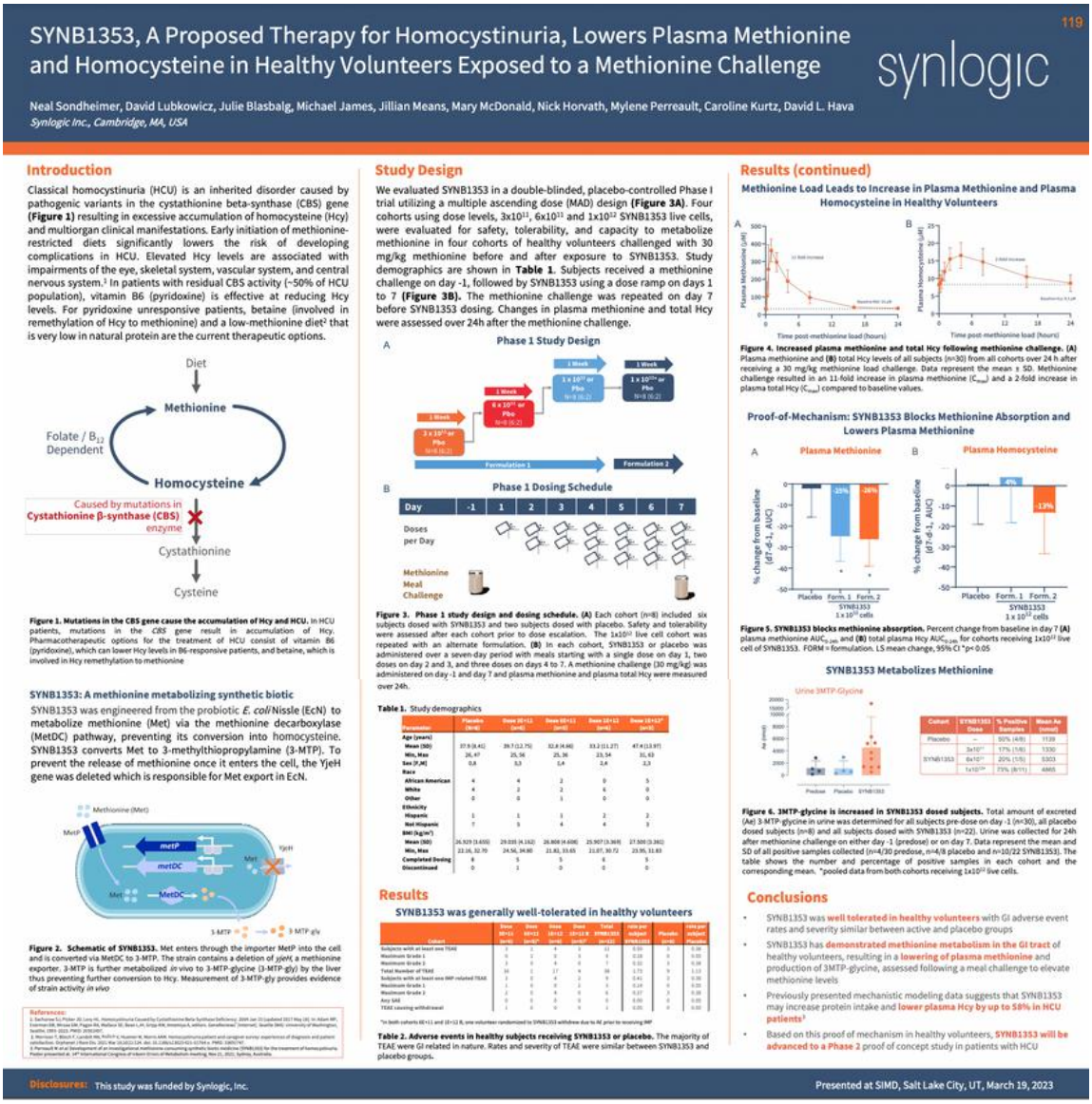
INDUSTRY NEWS

Synlogic Announces Data Presentations at the Society for Inherited Metabolic Disorders (SIMD) 44th Annual Meeting

CAMBRIDGE, Mass., March 20, 2023 (GLOBE NEWSWIRE) -- Synlogic, Inc. (Nasdaq: SYBX), a biotech company that is advancing therapeutics based on synthetic biology, announced that positive data from the company's Phase 2 Synpheny-1 study for phenylketonuria (PKU) were presented in a podium presentation yesterday during the Society for Inherited Metabolic Disorders (SIMD) 44th Annual Meeting in Salt Lake City, Utah. In separate poster presentations, the company also presented clinical data and preclinical data related to its homocystinuria (HCU) program.

Phase 1 results show that SYNB1353 was well tolerated in healthy volunteers, who were given a methionine load before being given the trial drug. In addition to being safe, the drug demonstrated proof of concept by lowering plasma methionine. Previously presented mechanistic modeling data suggests that SYNB1353 may increase protein intake and lower plasma Hcyby up to 58% in HCU patients. Based upon this proof of mechanism in healthy volunteers, SYNB1353 will advance to Phase 2 proof of concept study in patients with HCU.

To read the full press release, visit http://bit.ly/3JAgGtT
To download the poster, visit https://bit.ly/3K2ZmPJ



Travere Therapeutics to Present Abstracts at the Society for Inherited Metabolic Disorders 44th Annual Meeting and the American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting 2023

SAN DIEGO, March 14, 2023 (GLOBE NEWSWIRE) -- Travere Therapeutics, Inc. (NASDAQ: TVTX) announced that the Company will present a quantitative systems pharmacology (QSP) model for predicting the effects of the investigational enzyme replacement therapy pegtibatinase, in classical homocystinuria (HCU), as well as real-world evidence on the prevalence and potential underdiagnosis and/or underreporting of HCU in the United States, at the 2023 Society for Inherited Metabolic Disorders (SIMD) 44th Annual Meeting in Salt Lake City, UT, March 18-21, 2023. The Company and its collaborators will also provide presentations from the Company's ongoing longitudinal natural history study of people living with HCU, and HCU incidence estimates based on gnomAD database evaluation at the American College of Medical Genetics and Genomics (ACMG) Annual Clinical Genetics Meeting in Salt Lake City, UT, March 14-18, 2023.

To read the full press release, visit http://bit.ly/3K547YP

Summary of SIMD 2023 posters from Travere Therapeutics SS

Development of A Patient Identification Algorithm to Estimate Prevalence of Homocystinuria (HCU) in the United States (US)

The poster, Development of A Patient Identification Algorithm to Estimate Prevalence of Homocystinuria (HCU) in the United States (US) shared at the 2023 SIMD conference showed that the prevalence estimates vary widely based upon the approach and cohort definitions. A large portion of patients with high total homocysteine levels and clinical presentations indicative of HCU did not have a corresponding diagnosis of HCU, suggesting potential underdiagnosis and/or underreporting.

A Quantitative Systems Pharmacology (QSP) Model for Classical Homocystinuria Predicting Efficacy of Treatment

Travere's QSP computational model successfully illustrated the complex interactions that exist between metabolites of the methionine cycle and factors such as dietary methionine restriction and use of therapies such as Betaine and pegtibatinase. The model recapitulates and extends the same conclusions as numerous preclinical studies, predicting tHCY lowering associated with pegitbatinase use in addition to or independent of dietary protein restriction and betaine supplementation.

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

SURVEY PARTICIPANTS NEEDED!

Attention patients & caregivers of ***Classical HCU***, ***Cobalamin Disorders***, & ***Severe MTHFR***!

You are invited!

Petri Bio is working on a potential solution for HCU. We are trying to better understand the specific needs of those with HCU and the families that care for them. Please click through this 1-min survey to provide your feedback and join the sign-up list for our future products!

If you...

- Have been diagnosed with Homocystinuria (HCU) or another rare metabolic disorder
- Are a caregiver for someone who has been diagnosed with HCU or another rare metabolic disorder
- A researcher/healthcare specialist in the field of HCU or other metabolic disorders

you can also scan the QR code to join

We want to hear from you!

Join Now

More information

https://petribio.com/
info@petribio.com

Click here to take the brief survey!

UPDATE: AMAZON ENDS AMAZONSMILE PROGRAM

Global Genes
Allies in Rare Disease

End of AmazonSmile program impacts non-profits

February 14, 2023

Last month, Amazon announced it is sunsetting its AmazonSmile program on Monday, February 20, 2023. The program was started in 2013 to benefit non-profits of all sizes by allowing anyone purchasing goods through the Amazon site to allocate a small percentage of each purchase to the non-profit of their choice. For the past decade, these donations have allowed non-profits to add anywhere from a few hundred dollars to thousand dollars to their bottom line, and as anyone in the non-profit industry can tell you, every penny counts.

Read the full blog post & letter sent to Amazon CEOs by Global Genes and Members of the RARE Global Advocacy Alliance here

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UPDATE

HCU Network America has now partnered with iGive!

Shopping online @ any of 2,000 stores like Walmart, Travelocity, Overstock, & MANY more, means a donation to us!

HCU NETWORK AMERICA

Join iGive now using the link below!
https://iGive.com/RJeOWr

Need help setting up your account?
Visit
https://hcunetworkamerica.org/donate/igive/

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

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