The HCU Herald

Featuring...

HCU Hero
Mason from Wyoming

HCU NETWORK AMERICA

March 2023

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

HCU HERO: MASON FROM WYOMING

In the fall of 2014, we were living the classic “American dream”. We had a 2-year-old daughter, a yellow Labrador and were about to have our son. Mason was born in early December 2014 in Evanston, Wyoming and everything was going “normal”. He had a little bit of nystagmus (involuntary, rapid and repetitive movement of the eyes) and was a little tired after his circumcision, but the doctors and nurses said everything was fine and to go home and enjoy our healthy baby boy. Little did we know, we were about to become fully aware of the world of rare metabolic diseases.

On Mason’s 6th day of life, we were just hanging out at home and enjoying our new addition to the family when we got a call from the Children’s Hospital in Colorado. They had the results of Mason’s newborn blood screening and his levels were not where they should be. They said that it is very likely that he has a metabolic disease and they recommended we contact his pediatrician to get additional labs done that day to know the next step we would need to take. The visit with the pediatrician was a whirlwind. They were fairly confident that he had a metabolic disease but didn’t really know which one. They informed us of a few worst-case scenarios to attempt to mentally prepare us for the worst. When we left the doctor’s office, we sat in the car trying to figure out what to do next. We were still overwhelmed by the whirlwind of information we received and the unknowns of which metabolic disease and associated complications he may have. As we sat there Shandra was trying to feed Mason and he wouldn’t latch on. The next thing we knew he was as limp as a noodle; completely lethargic.

HCU HERO: MASON FROM WYOMING

We marched right back into the doctor's office for help. Calls were made to the Children's Hospital and as a group, we decided the best thing to do was get Mason to Colorado. In a rushed panic we got our belongings together and had friends take care of our house and our dog. Shandra jumped in the life flight airplane with Mason, and Brandon took Madyson and made the 6-hour drive in the dark in December to Denver. Looking back, we did have one thing on our side: the weather and roads were in amazing shape for that time of year.

We spent the next 9 days in the NICU at Children's in Colorado learning more than we ever expected to know about Cobalamin C deficiency. It was a lot to take in, but they were able to break down Mason's condition into terms that we could understand. They ran several tests to understand his current metabolic state as well as a thorough examination of his eyes and his heart. Cobalamin C has several unpredictable side effects, but one that is common with Cobalamin C deficiency is vision loss. During this initial visit Mason did not show any signs of vision loss.

> **"They made sure that we were comfortable giving Masen his hydroxocobalamin shot everyday. In all openness, it was an intimidating task, but we knew he needed it to live."**

We met some amazing doctors and nurses at Children's and everyone took great care of us. They helped us establish contacts with all the pharmacies for Mason's unique medications, which is quite an ordeal for the cystadane and compounded hydroxocobalamin. They also made sure that we were comfortable giving Mason his hydroxocobalamin shot everyday. In all openness, it was an intimidating task, but we knew he needed it to live so we cowboy'd up and learned how to do it. After his levels were in reasonable shape and they were comfortable that he had all the medications he'd need, they let us go home.

HCU HERO: MASON FROM WYOMING

Once we got to our home in Green River, WY we had to figure out our new complex life, which included keeping a watchful eye on our son for potential seizures, lethargy, and the other issues the doctors had warned us about. We were very paranoid about missing one of Mason's critical doses of his medicine so we built a checklist to make sure we didn't miss any doses. We also had to establish a schedule to get Mason to the local hospital so that he could get his blood drawn for lab work every week. Reflecting on all of this now, we feel we did tremendously well. Mason is now 7 years old and he hasn't missed a single dose of his critical medicines.

Since we were so far from the Children's Hospital we couldn't visit them every week per the normal protocol, so instead we would go down once a month. Mason's first visit back to Children's would be one that we would never forget. At the end of his eye exam, Dr. McCourt told us that she was starting to see the center of Mason's retinas beginning to die - atrophy and the beginning stages of macular degeneration. Dr. McCourt was amazing through this traumatic time: explaining what she was seeing in terms we could understand, referring us to different resources and most importantly giving us time to deal with this news. We spent the next 45 minutes sitting in the exam room crying our eyes out. Once we gathered ourselves, we thanked Dr. McCourt for her kindness and we headed out.

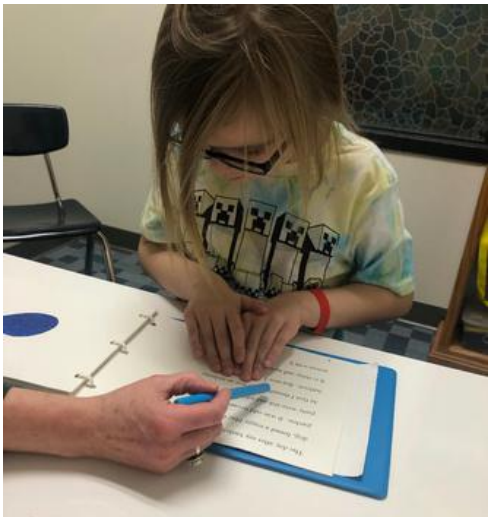
We continued to make our monthly trips to Colorado so that the doctors could continue to monitor Mason. At 6, 12, and 18 months we had longer visits where Mason was sedated and they performed MRIs for monitoring. Through the checkups Mason continued to have high chemistry for a normal individual, but they were on the lower side that the doctors had seen for a Cobalamin C patient. It felt good that the hard work and diligence to make sure he got all of his medications was paying off. The doctors were impressed and completely baffled that the dead spots on his retinas were continuing to grow with good metabolic chemistry. Dr. Weisfeld-Adams (metabolic) and Dr. McCourt (ophthalmology) were determined to figure out what was causing the vision loss and we promised to help them out in any way that we could. From then, Mason's vision has remained relatively stable since he was about two and a half years old. Dr. McCourt continues to closely monitor to make sure that the dead spots in the center of his eyes do not grow and closely monitors his peripheral vision for any signs of loss.

Unfortunately, cancer took Dr. Weisfeld-Adams away from us in 2018. We were heartbroken that he is no longer here, but we still have amazing metabolic doctors that take care of Mason. We were honored to be invited to his funeral. Brandon was able to speak during his services and let his close family and friends know how special he was to our family and how he was able to make this extremely difficult situation we were living with endurable.

As Mason has grown up, we have done our best to let him try anything and never hold him back. He has played indoor soccer and participated in the local baseball "Challenger League". Soccer is getting a little tougher for him as the kids are getting faster and more aggressive. He has fallen after bumping into the kids and he gets embarrassed, but we encourage him to get up and shake it off and keep playing. The "Challenger League" is a phenomenal experience for Mason. It is a league that is available for anyone with special needs. It is split up into two groups, 13 years and younger and 13 years and older. There are no real rules, they just let every kid have a chance to bat every inning and everyone has a blast. Kids from the "minor" and "major" leagues will volunteer and help the kids field the ball and run the bases. It is awesome to see the kids' faces light up after they hit the ball and take off for first base. Mason especially likes to be the last batter because that player gets to clear the bases with an automatic home run!

Mason has recently joined Cub Scouts and has had a blast doing it. He's learning a lot about helping his community. His Tiger pack had a fundraiser and built birthday packages for the local food bank. Through this effort, they gathered 362 pounds of food from the community to give birthday cake supplies to those in need! Mason also had a lot of fun with the pinewood derby and took 1st place for the Tiger's class at the district races.

Having a vision impaired student in the Wyoming schools has been a challenge. Not because they don't want to help, but there are few vision impaired people in our rural state. There are not many providers and few are familiar with the specific needs of vision impaired students.



Mason's first IEP with his preschool was the most challenging. The short story is they were not providing the vision services that he needed and that we agreed to in the IEP, so we ultimately had to file a complaint with the state. It was a tough process to go through and we spent hours researching special needs education to ensure that we knew what we were talking about as we fought for Mason's rights. We can't stress enough that you must be the biggest advocate for your child; no one else will advocate as hard as you will. You need to educate yourself to understand the special education laws and your child's rights so that they can have the best education that they can get.

Mason is going into the 2nd grade this year and the elementary school experience has been better than the preschool experience. We stole an idea from Mason's TVI (a teacher of students with visual impairment) that also has a son with vision loss. We created a flier to help introduce Mason to the school staff and understand his situation. We included pictures, his favorite things, and critical parts of his special needs in simple terms. This was a huge hit, and all of the staff truly cares about him and looks out for him. They've made sure that the kids don't leave things out in the hallway and even added high-contrast material to the steps and many other simple things to make sure he's safe at school. In kindergarten, Mason learned all the letters of the alphabet in print and in Braille, which completely blew us away. He's also doing better with his orientation and mobility/cane skills. On October 15th, White Cane Day, Mason's Orientation and Mobility Specialist set up activities for his kindergarten class and the other grades to let the other children understand what it's like to utilize a white cane.

Overall, our life has taken us down a path we never thought we would go on, but we wouldn't change it for anything. We've met fantastic people and lifelong friends through the OAA conferences, through our trips to the Children's Hospital, and through the schools and service providers. If we were to leave you with our biggest learnings on this wild ride known as life, it would be:

- Leverage other people that are going through the same things that you are via organizations like the OAA. You are not alone and people going through similar situations want to help.
- Understand the laws and your rights with respect to special education. No one will be a bigger advocate for your child than you. Education is the best tool you can have.
- The greatest quote that we've ever heard is "Life is a journey, not a destination." by Ralph Waldo Emerson. You have to make sure that you take time to enjoy those little precious moments in life when your child is in a pure state of joy or does something you never thought they would be able to, like riding a bike!

To read Mason's story on our website, visit
https://hcunetworkamerica.org/cobalamin-patient-stories/mason/

OPPORTUNITY ANNOUNCEMENT

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!

Vegetable Pancakes

Makes 3 pancakes: Each pancake is 0.6g protein

Ingredients:

- 3/4 c Shredded Green Cabbage
- 1/4 carrot(s) Shredded Raw Carrots
- 1/4 c Chopped Scallions
- 1 c Cambrooke MIxQuick Baking Mix
- 1/2 tsp Salt
- 1/4 tsp Ground Black Pepper
- 1 tsp Grated Ginger Root
- 1 TBSP Bragg's Coconut Aminos
- 1 c Water

Notes:
*You can add your choice of veggies. Make sure they are shredded or chopped small for even cooking.

*To save time, you can use 1 cup coleslaw mix for the pancake in place of shredding cabbage and carrots.

Directions:

1. Preheat 2 tablespoons of vegetable oil in a small skillet over medium-high heat.

2. Combine the MixQuick, ginger, coconut aminos, and water in a medium bowl. Whisk until combined. It should look like pancake batter. Add the cabbage, carrots, and green onions and gently fold with a spatula to mix.

3. Add 1/2 cup of the batter mixture to heated pan. Spread thin with a spatula. You want the pancake to be thin. Cook until lightly browned, about 4 to 5 minutes and flip. Cook on this side until lightly browned. Remove from skillet. Repeat for the rest of the batter. Cut into pieces and serve warm with your choice of sauce.

March 2023, HCU & You: Ask Methia

Dear Methia,

Help! I feel guilty that I can't give my child what their sibling is having.

I have three awesome kids. My older two children do not have any food allergies or diet restrictions. My youngest has HCU, and has been on treatment with formula and a low protein diet since he was ten days old. For the first few years of life, it was easier to conceal the differences in their diet so that my youngest didn't notice. However, now that he is in preschool and being exposed to the foods/snacks that other children eat, he is asking more questions. He is beginning to express to us that it's "not fair" that his siblings can eat "normal" chicken nuggets when he eats Tweekz, and that his friends can have "normal" ice cream but he has to have sorbet or dairy free. I feel so guilty about this, and really just want him to feel satisfied with his diet. How can I help him?

Sincerely,
"Monkey in the Middle"

Dear Monkey,

Food plays such a big part of our day-to-day lives, and this makes following any diet challenging. Low protein diets that are medically necessary are even MORE challenging for a couple of reasons. First, protein is in (almost) everything, and lower protein retail options are relatively new to the market. Second, the consequences of going off diet are much higher than they are for, say, someone embarking on a weight loss journey. This adds an emotional component to dietary compliance that is even more heightened for children, who have BIG emotions! I hope you already know that you are a great parent for seeking ways to help your child. Here are a few suggestions:

- **Prioritize meals that "everyone eats."** This can be hard if your older children are picky, but there are ways to make dinners such as "Taco Night" more inclusive. For example, nix the ground beef for the rest of the family and offer EVERYONE jackfruit tacos. Substitute red beans and rice for cauliflower rice, a lower protein side that everyone can enjoy (and reap the health benefits of a lower calorie, higher fiber option!). Don't worry - most people eat way more protein than they actually need. Cutting back a little won't make you deficient!

- **Encourage all of your children to help with low protein meal preparation.** If everyone is eating the same thing, then everyone (not just your child with HCU!) will begin to normalize these lower protein meals. Being a part of the process is a fun way for all of your kids to bond, and your child with HCU will feel like their siblings are making the effort to incorporate the low protein diet into their lives.

- **Come up with a meal schedule and snack solutions that work well for your family.** Maybe you decide as a family to only keep low protein snacks in the house, and that higher protein snacks can be purchased at school. Perhaps "Pizza Night" is once every two weeks, and you offer both standard protein and low protein options at this specific meal. Reach an understanding with everyone in the family so that your child with HCU isn't let down or startled when something "off limits" to them is offered.

It's so important to continue to "make food fun," despite many restrictions. Fortunately, with so many new gluten free and vegan food options that are naturally low in protein, finding foods that the entire family can enjoy is becoming easier! Ask your metabolic dietitian for some suggestions and updates on the newest products.

Sincerely,
Methia

New Resource

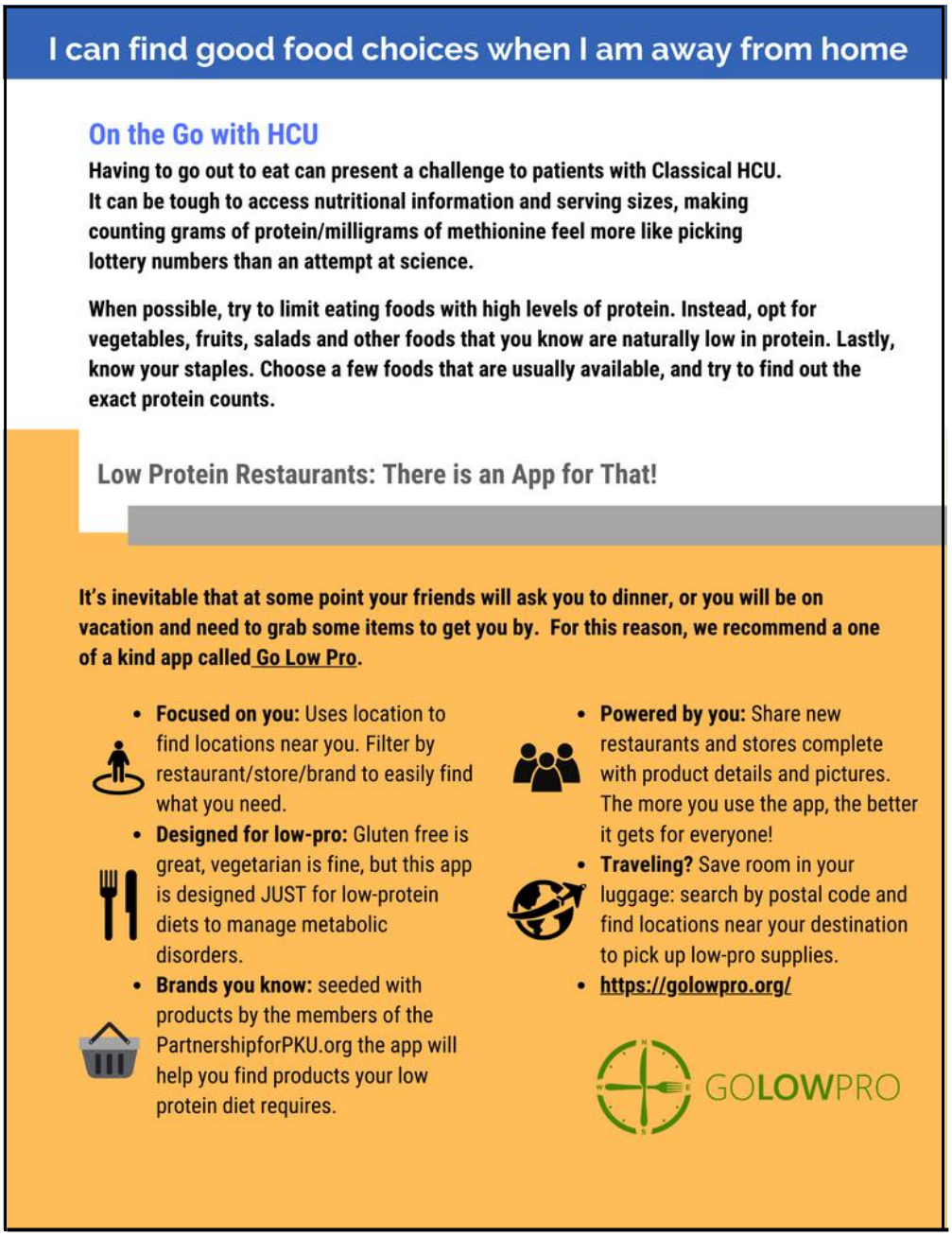
Let's help you *Transition to Adulthood*!

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

I can find good food choices when I am away from home.

CLICK HERE

to download this resource



I can find good food choices when I am away from home

On the Go with HCU

Having to go out to eat can present a challenge to patients with Classical HCU. It can be tough to access nutritional information and serving sizes, making counting grams of protein/milligrams of methionine feel more like picking lottery numbers than an attempt at science.

When possible, try to limit eating foods with high levels of protein. Instead, opt for vegetables, fruits, salads and other foods that you know are naturally low in protein. Lastly, know your staples. Choose a few foods that are usually available, and try to find out the exact protein counts.

Low Protein Restaurants: There is an App for That!

It's inevitable that at some point your friends will ask you to dinner, or you will be on vacation and need to grab some items to get you by. For this reason, we recommend a one of a kind app called Go Low Pro.

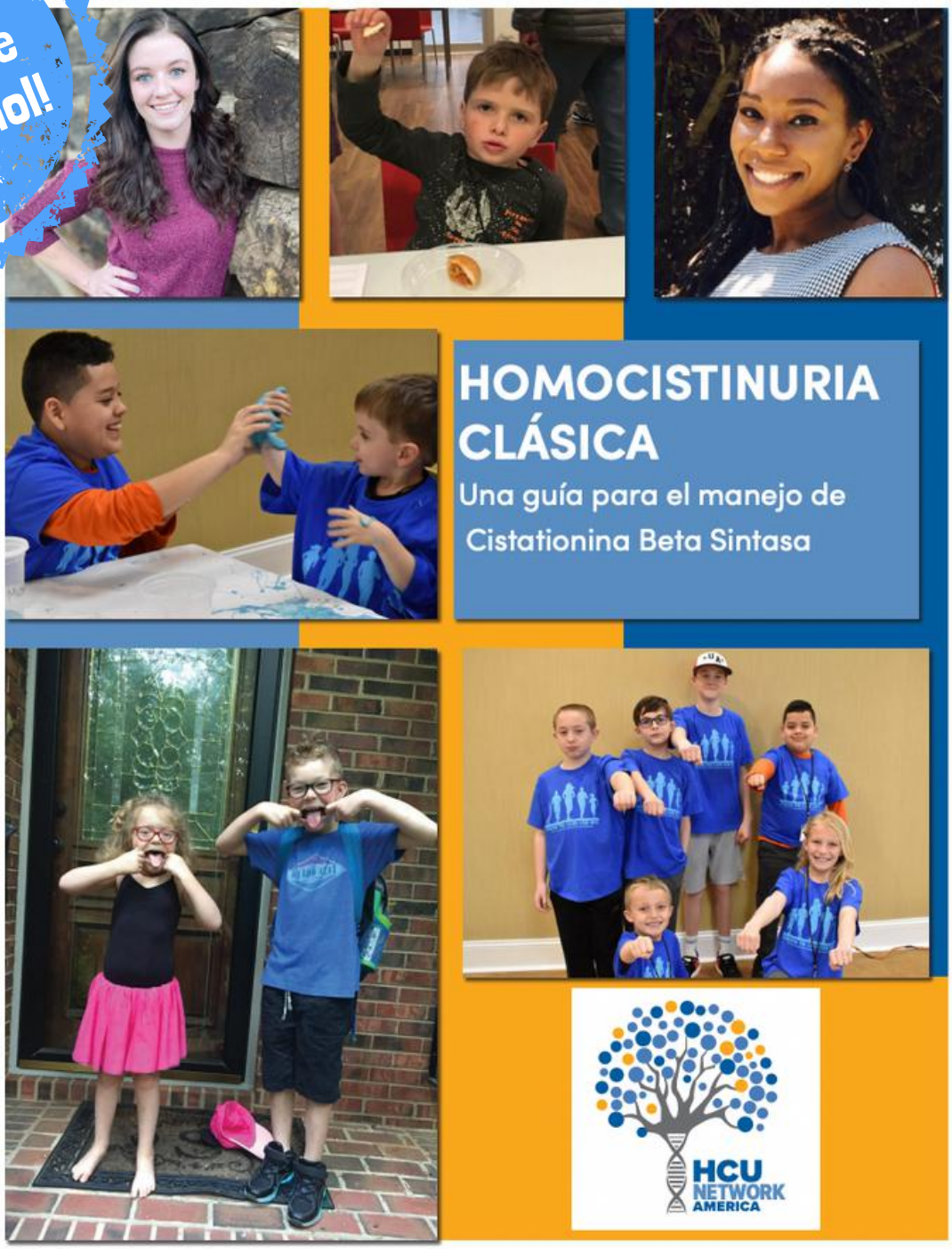
- **Focused on you:** Uses location to find locations near you. Filter by restaurant/store/brand to easily find what you need.
- **Designed for low-pro:** Gluten free is great, vegetarian is fine, but this app is designed JUST for low-protein diets to manage metabolic disorders.
- **Brands you know:** seeded with products by the members of the PartnershipforPKU.org the app will help you find products your low protein diet requires.
- **Powered by you:** Share new restaurants and stores complete with product details and pictures. The more you use the app, the better it gets for everyone!
- **Traveling?** Save room in your luggage: search by postal code and find locations near your destination to pick up low-pro supplies.
- **https://golowpro.org/**

GOLOWPRO

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

New Resource

¡Ahora disponible en español!



Esta guía está diseñada para brindarle una descripción general de qué es la homocistinuria, cómo se trata y cómo manejar mejor la dieta y el trastorno.

Haga clic aquí para descargar la guía

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

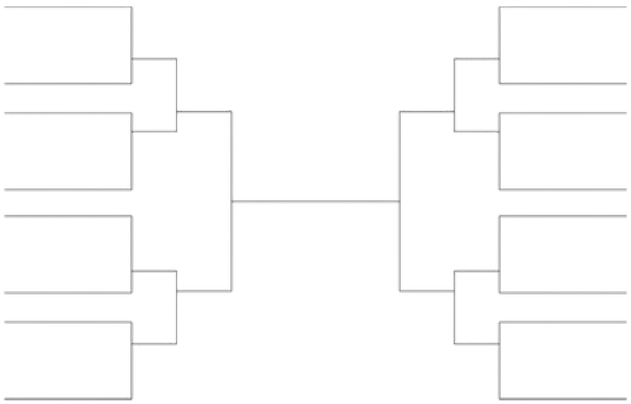
Join iGive now using the link below!
https://iGive.com/RJeOWr

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

MARCH FUNDRAISING EVENT: MARCH MADNESS

March Madness Selection *Sunday 2023 is* ***March 12***!
Start assembling your pool & raise funding for resources, tools & outreach for **HCU Network America!**

March Madness is a three week period packed full of buzzer beaters and is a sports poolers dream. But how can you keep the excitement alive when most of your members have had their brackets busted? A great option is our Madness Squares pool format, as every game of the tournament will have a winner!



How Do Madness Squares Work?
If you are familiar with Super Bowl Squares, the main idea is the same for March Madness. A 10x10 grid of boxes is setup and each row and column is given a number from 0 to 9. Just like in Super Bowl Squares, each square of the grid can be claimed by a pool member.

Winner breakdown
Each round is worth a set number of points. You can determine this on your own, but be sure to let all of the entries know before the tournament begins what the scoring system will be. (You should write the point values under each round at the top of the bracket).

Declaring a Winner
Multiply the total number of correctly picked games in each round by the points assigned to that particular round. Tally all rounds together and the person with the highest point total wins!

- For further instructions and to print your bracket, visit: https://www.printyourbrackets.com/howtomarchmadness.html
- For online tools, check out: https://www.runyourpool.com/march-madness-squares-pools.cfm

EVENT ANNOUNCEMENT

Family Camp, hosted by PKU Alliance

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 !

UPCOMING EVENTS

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

Classical HCU Parent-Caregiver Meetup

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

Saturday, 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

Low Protein Cooking Class

April 1 | Time: 2 PM EDT

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

Clinical Trial for Classical Homocystinuria

Sponsor: Aeglea BioTherapeutics ● **Drug name:** Pegtarviliase. ● **Therapy type:** Enzyme therapy

Administration method: Subcutaneous injection ● **Goal:** Reduction of total plasma homocysteine (tHcy)

Phase 1 | Phase 2 | Phase 3 | Phase 4

The purpose of this **Phase 1/2 study** is to evaluate the safety, pharmacokinetics and pharmacodynamics of pegtarviliase in subjects with homocystinuria due to CBS deficiency.

Actively recruiting

DURATION	AGE OF PARTICIPANTS	TO QUALIFY
Up to 14 weeks, to include a screening period, 4 weeks of treatment (1 dose/week), and a follow-up period	≥12 years of age in UK (and select study sites in Australia) ≥18 years in US and Australia	Plasma tHcy ≥50 µM (rounded to the nearest whole number) and documentation of previous tHcy ≥80 µM

**The study details above may change. For the most current information and to see additional eligibility criteria, please visit: https://clinicaltrials.gov/ct2/show/NCT05154890*

Trial sites

Dallas, TX
UT Southwestern Medical Center

Birmingham, United Kingdom
University Hospitals Birmingham NHS

London, United Kingdom
Great Ormond Street Hospital for Children
Guy's and St Thomas' Hospital NHS Foundation Trust
University College London

Salford, United Kingdom
Salford Royal NHS Foundation Trust

Australia, New South Wales
Westmead Hospital

Australia, Victoria
Royal Children's Hospital
Royal Melbourne Hospital

☑ **Some study visits may be done at your home.**

☑ **Study-related expenses** *(travel service, reimbursement for loss of earnings, and other study-related expenses)* **may be provided.**

Contact: Study site OR Aeglea Clinical Department 1.855.509.9921 clinicaltrials@aeglea.com

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

PAID OPPORTUNITY

You're Invited!

Your feedback is important.

CLASSICAL HCU TREATMENT DEVELOPMENT

Market Research Study

We are seeking individuals to participate in a market research study discussing their experience living with classical homocystinuria and their interest in a new medical treatment that is currently being studied.

If you:

- live in the U.S. *(some exceptions may apply)*

AND

- have been diagnosed with **classical homocystinuria**

OR

are a **caregiver** for someone who has been diagnosed with classical homocystinuria

AND

- are **18 years or older**

you may qualify to participate in a Zoom interview study. All information and responses during this interview will remain confidential.

If you qualify and complete a 60-minute interview, you will be **compensated $100 for your time and participation**. Interviews are being run until **March 9.**

Interested?

To see if you qualify, please fill out the following form and a representative from ClearView will be in touch!

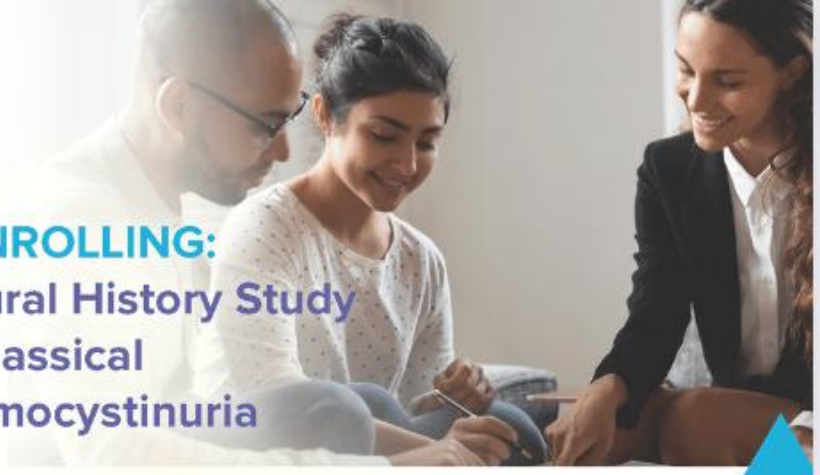
We'd like to hear about your current classical HCU management plan and level of interest in a new treatment in development.

Example Topics:

- **What medical or lifestyle treatments have you been prescribed for classical HCU?**
- **How satisfied are you with your current treatment plan?**
- **If a new treatment became available for classical HCU, what questions would you have?**
- **What treatment dosing and device factors would you consider before switching to a new treatment?**
- **What medical or lifestyle goals are you looking to achieve from a new treatment that you have not been able to achieve from your current treatments, if any?**

Please note, access to a laptop or tablet will be necessary to complete the interview as we will ask you to look over a brief description of the new treatment

Click here to apply!



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

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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

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

Attention friends in Canada!

This message is for you.

https://canpku.org/

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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