The HCU Herald

Featuring...

HCU Hero
Cam from Illinois

2023

January 2023

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

HCU HERO: CAM FROM ILLINOIS

Our child's name is Cameron (mostly known as Cam), and she is six years old. Cam was born in August of 2016 at 38 weeks through cesarean section due to being breached, but otherwise, it was an overall healthy "normal" pregnancy. Thanks to the newborn screening tests, Cameron was diagnosed at birth with her disease. They called us within the first two weeks of bringing home our newborn baby from the hospital saying that she had tested positive for a genetic disease: Methylmalonic Acidemia, which was caused by Cobalamin C deficiency.

After multiple Ophthalmologist appointments and a sedated imaging scan through neurology, Cam received a diagnosis of retinal degeneration in 2019. As you can imagine, we as her parents took this news hard. We feared the unknown. We wondered what this diagnosis might mean for our baby's quality of life, her future and for us as a family. After our initial feelings and emotions set in, we knew we needed to make some mental adjustments so that we could be at our best to provide for our special baby and be her advocates.

HCU HERO: CAM FROM ILLINOIS

Our friends and family have been great. This journey has humbled us all and shed light on so many things we never imagined we'd face or ever go through as we love and support Cameron. Cameron has touched many of our loved ones and has a special place in their hearts. We rallied together in 2020 for World Rare Disease Day and hosted a fundraiser to raise money and donate to help with resources and research for all genetic diseases. The love and support we received was amazing - all for the love of our sweet Cam. Her journey is teaching us all not only knowledge about this rare disease but empathy for all those affected.

> "Her disease affects her body's strength and energy but her motor skills have grown tremendously, and she is so very active and full of life."

Although Cameron has developmental delays and struggles with vision impairment she continues to thrive and amaze us in other aspects of her life. She is bilingual, speaking both English and Spanish fluently; the first in the last two generations on her mom's side to do so. Her disease affects her body's strength and energy but her motor skills have grown tremendously, and she is so very active and full of life. She is progressing in her understanding of situations and how to react to them. Cameron also amazes us with her remarkable memory! If you've ever met her, taken her somewhere, played a song for her or simply spent time with her, she will never forget it and will remind you any time she sees you again.

The biggest challenge as her parents honestly comes from just feeling helpless at times, wanting to give Cameron all the treatment and help she needs to be able to do everything a child her age should be able to. As of now, Cameron's biggest obstacle seems to be her loss of vision. She has struggled with the lack of eyesight from the very beginning, has seen different ophthalmologists, and is dealing with retinal degeneration. It is sometimes challenging for us to determine in certain situations if Cameron is not comprehending something or if her eyesight is rendering things. I think simply having patience and grace for her as she learns to navigate through life is how we will overcome these situations.

If we could give newly diagnosed families advice it would simply be this - the promise that no matter how it seems right now, everything will be ok. Many emotions will come into play initially and I hope you know that your feelings are valid, and that you are not alone on this journey. Time will bring you understanding, knowledge, hope and most importantly strength. Be patient with your child and yourself as you're figuring out this way of life together. Don't measure your child against others or what medical standards say. Don't dwell on the setbacks and celebrate the wins, big or little. Enjoy your little one because time flies by and you are truly blessed to be given the honor of watching and helping this amazing being grow. Stay strong, stay together and...
remember how lucky you are to love someone RARE!

This HCU Hero story was written with love by Sarah, mom of Cam.

Cannoli Icebox Cake

***Yields 4 servings* | Serving Size: 87.5 g | Protein per serving: 1.9 g**

Ingredients:

- 1/2 c Tofutti Better Than Ricotta
- 1/2 c Powdered Sugar
- 4 TBSP Violife Just Like Cream Cheese Original, softened
- 1/4 tsp Almond Extract
- 3/4 c Cool Whip, divided
- 5 square(s) Honey Maid Grahams, Honey
- 2 TBSP Semi-Sweet Chocolate Mini Chips

Directions:

1. In a medium bowl add the ricotta, powdered sugar, softened cream cheese, and almond extract. Using a spoon or small spatula mix all ingredients until smooth and well combined. Fold in 1/4 cup of cool whip into the cheese filling.
2. In a small loaf pan, (6.1"(L) x 3.4"(W) x 1.9"(H)), spread a small amount in the bottom. Place one of the graham cracker squares in the bottom. You can break it to fit, as you will want the bottom fully covered with the crackers. Top with half the cheese filling. Sprinkle with one tablespoon of mini chocolate chips. Place another graham cracker square on top of cheese layer. Top with other half of the filling and sprinkle with 1 tablespoon mini chocolate chips. Top with final layer of graham cracker squares. Spread the 1/2 cup of cool whip on top.
3. Cover the pan with a lid or plastic wrap and refrigerate for at least one hour or up to overnight. You will want it to sit of a while to allow the filling to soften the graham crackers. Serve chilled.

2022 $30k Match Challenge Highlights

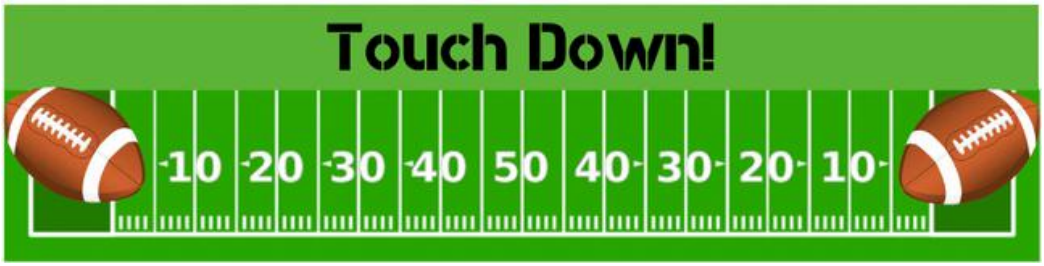
Our annual appeal started in October with HCU Awareness month. We had a good start to the month with several fundraisers hosted on behalf of HCU Network America. In October we raised $2,344 thanks to the 2 families who set up fundraisers on behalf of the organization and to the many others for their individual donations.

In November we had another wonderful opportunity to raise awareness and funds for HCU Network America with Thanksgiving and Giving Tuesday. We also sent an email with an appeal letter that those in the community could share with their family, friends, co-workers and others. By the first part of December, *we were 30% of the way to our goal.* Thank you to those who set up #GivingTuesday fundraisers.

By mid-December, as our annual appeal was about to come to a close, we sent out a few more emails recapping our accomplishments of the past year. Because of our successful campaign, we were able to accomplish a lot in 2022! With these emails, we were able to take in $13,979.52, with a final total of $22,573 to continue our mission in 2023!

While we did not meet our financial goal, we are still very thankful for the generous donations from our supporters! We couldn't have had a successful 2022

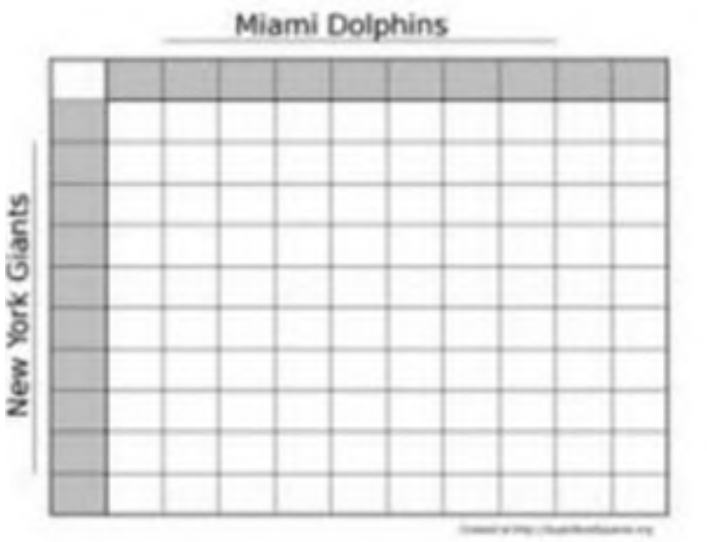
JANUARY FUNDRAISING EVENT: SUPER BOWL SQUARES



Big sporting events are a great way to not just show your team spirit, but they are a fantastic way to **raise funds for your charity of choice - HCU Network America!**

How it works:

The cost is $5 or $10 donation per square. The purchaser writes his or her name in the suqare, and then the seller will randomly pick numbers 0-9 from each team in the game, and assign that number to a particular row or column (the grey shaded columns). These numbers represent the last numbers in the score of each team at the end of the game. In other words, if the final score is Giants 17 - Dolphins 14, then the winning square is the one with a Giants number 7 and a Dolphins number of 4. Since no one knows what numbers each square will represent, the odds are the same for everyone. So good luck and have fun!



Further instructions and templates:
https://www.printyourbrackets.com/nflweekly100squares.html

Online tools:
http://footballsquaresonline.com

https://www.runyourpool.com/nfl-football-pools.cfm

Winner Breakdown:

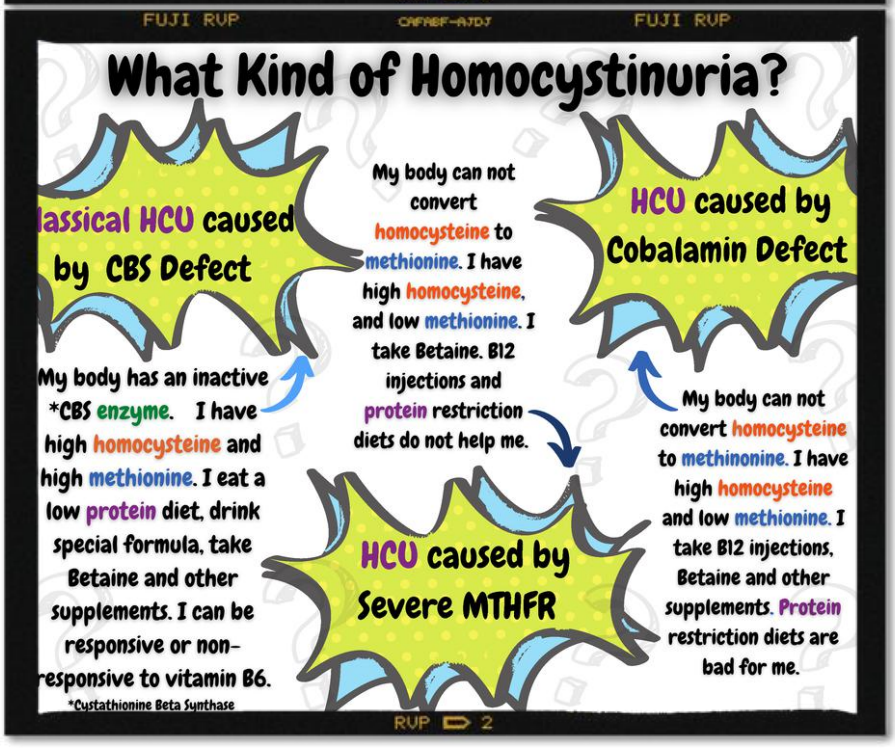
If you sell 100 squares (you must sell all squares to host the fundraiser) at $10 each, the funds raised are $1,000. You can chose to split the pot evenly and the winner will get $500 or get a large item donated for the winner and keep all funds raised toward your fundraising effort. (You can also decide to split the prize money and give some at half time or the end of each quarter.

Need help hosting a fundraiser? Email us - info@hcunetworkamerica.org

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 our first resources to go along with it!



The first two objectives in the guide are: I know what type of homocystinuria I have, and I can explain the basics of my condition.

Knowing the type of homocystinuria you have and the basics will allow you to better advocate for yourself and make sure your treatment aligns with your specific type of HCU.

The first infographic helps provide you with a simple explanation of the vocabulary you need to understand the basics. The vocabulary boxes are color coded to match the words on the second infographic.

The second infographic breaks down the three main types of homocystinuria, what their body can't convert, and what their treatment may look like.

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

The New Year isn't the only thing we're celebrating!

Our **Rare-X Data Collection Program** now has representation in 19 states....and counting!

Be counted!

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

REGISTER NOW!

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

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.

Registration for Rare Disease on Capitol Hill 2023 will open on January 4th, 2023.

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

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

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

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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

Live better, together!

Contact Register

What is the contact register?

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

What will this information be used for?

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

How do I participate?

The contact register form takes approximately 3-5 minutes to complete. You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to: https://bit.ly/3OJuF1W