

The HCU

Herald



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HCU HERO: SUMMER FROM CONNECTICUT

Our journey with Homocystinuria started in October 2015 standing in our pediatrician's office glaring at my daughter's extended limbs and listening to her heart murmur. "I think she has Marfans", the doctor said alarmingly followed by "You need to see a cardiologist right away". At the cardiologist's office, I pondered and searched the devastating effects of Marfans. Thankfully, her cardiologist visit was somewhat successful since Marfans had been ruled out, but what was this then?



We were referred to Dr. Wendy Chung, Clinical Geneticist at Morgan Stanley Children's Hospital in NYC. Our visit was simple and informative. Blood samples were taken from my husband, myself and our daughter, Summer. The wait was long for answers. In February 2016, Dr. Chung called me. "It's Homocystinuria", she said in a matter of fact tone. "You're going to have to come in for an appointment every three weeks for the first year", she said. Both my husband and I are carriers of the gene, but we don't have Homocystinuria. We then embarked on a lengthy journey which explained the strokes Summer suffered in utero, all the partial seizures she's had since birth, her difficulties in school and her rather tall, lanky physique from two short parents.

The first year was the longest. We went from protein packed dinners to abysmal tasting meals (we ate the same meals in support of Summer's new way of life.) We acquired 9 new doctors to deal with the complications and effects Homocystinuria has had on her body, not including her dental team or nutritionist. A big surprise was all the medicine and vitamins she would have to take regularly. After paying \$600 a month (after insurance) for Cysteine, the bigger shock was the cost.

Our biggest challenge with Homocystinuria is not the disorder itself, but making people understand the effects it has on my daughter's body and livelihood. How many school cooking classes and visits to her friends' homes will it take before the teachers and parents realize she can't eat chicken nuggets with a glass of milk followed by a huge serving of ice cream? Likewise, the learning challenges she's faced in school since the first day in Preschool have been monumental. Summer is in tenth grade now and they still haven't written her diagnosis of Homocystinuria in her IEP and fail to try and understand how it affects her learning as well as her physical self. As a family, we struggle to get her numbers under control. Since she wasn't diagnosed at birth and her palate had never been exposed to formula while an infant, Summer can't bring herself to drink any of it. We were thankful to find a Vitamin B6 in liquid form that she could tolerate (the vitamins and medicines tend to give her



terrible acid reflux).

Despite the physical challenges, learning difficulties and obstacles thrown her way, Summer has blossomed into a beautiful young lady we're very proud of. Our journey is never ending. However, we're grateful for those we've met along the way who are helping us along the path of our journey.



Email us your patient story! info@hcunetworkamerica.org

NEW RESOURCE



PARENT HANDBOOK Special Education Services

Special Education services are available for eligible students with disabilities. This handbook provides information about procedures, answers to frequently asked questions, and links to other resources. This information is meant to be an introduction to special education and may vary between states. For specific laws and regulations pertaining to special education in your state, visit your State Department of Education.



Check out this resource at:

<https://hcunetworkamerica.org/toolkits-and-checklist/>

COVID-19 Vaccination and HCU

December 28, 2020



What you need to know....

Hello,

As COVID-19 continues to impact all of our lives, there is some hope on the horizon with the introduction of several extremely effective vaccines from multiple sources. The speed in which these vaccines have been designed, tested and produced is truly amazing and due to the diligent work by many dedicated people. We will look back at this momentous effort as a true miracle of modern science.

Our next task, as a population, is to encourage and participate in widespread vaccination of the everyone in our population so that new COVID-19 infections will drop and we will be able to return to interacting with others in person outside our household. Only widespread vaccination, estimates are at least 50%, but more likely 70-90% of the population needs to be vaccinated, will protect us all.

According to science's current knowledge, having an organic acidemias (of any type) does not preclude getting the COVID19 vaccine and so we encourage everyone to get the vaccine when they are able. There will be some individuals who cannot get the vaccine (discuss with your own doctor if you have concerns), however having homocystinuria is not one of the reasons.

For those who have a protein restriction, neither of the COVID-19 vaccines being produced by Pfizer and Moderna contain any protein.

The Moderna and Pfizer COVID-19 vaccines are a very new kind of vaccine (for infectious diseases---the technology has been used to help treat cancer in the past). These vaccines are made of RNA (ribonucleic acid) protected by a coat of lipid (naturally occurring fats). RNA is the building block our own cells use as the code for producing important proteins. The cells read RNA code and build new proteins using amino acids already in your body. After the vaccine is injected into muscle, the muscle cells take up the RNA-lipid particles and strip the lipid coating off. The RNA code then teaches the muscle cells to produce a single protein from the COVID-19 virus. The code provided is the code for the COVID-19 spike protein, one protein that is found on the outside of the COVID-19 virus particle. The muscle cells will transiently produce the COVID-19 spike protein (but not the whole virus) for just a few days until the RNA code spontaneously degrades. The spike protein is 'presented' to the outside of the muscle cell wall where the immune cells circulating in the blood can see it. Since it is a foreign protein not normally found in our bodies, the immune cells recognize the spike protein as foreign and start gathering everything needed to destroy the protein. A second injection of the vaccine three weeks later really solidifies the immune response and commits it to the memory of the immune system. Thereafter, if the individual encounters the actual COVID-19 virus which is covered in spike proteins, her immune system will immediately recognize the foreign spike proteins and reactivate to destroy the virus. I have reviewed the data provided by Pfizer and Moderna to the FDA on the efficacy of their vaccine; it is remarkably effective.

Traditional vaccines, like the influenza vaccine, are based upon injecting the actual virus which has either been altered to not cause severe disease or has been killed. The vaccine still carries the viral proteins that the immune system will recognize as foreign. Traditional vaccines do contain protein, but the amount of protein injected is very small compared to the amount of protein consumed even in the most strict homocystinuria diet. Even though traditional vaccines contain protein, we still advise everyone with an organic acidemia to receive all recommended vaccinations.

To conclude, the Pfizer and Moderna COVID-19 vaccines do not contain any protein. They contain a code that will allow the transient production of a very small amount of a virus protein from amino acids already in the body. They will not affect methionine metabolism. It is crucial that all of us participate in the COVID-19 vaccination program in order to defeat this pandemic. I encourage everyone with homocystinuria to participate.

The CDC and your state's government's Board of Health have the most up-to-date information for vaccination plans in your area. Please be patient, much of the information about the tiering system that prioritizes vaccine delivery is on your state's government's Board of Health website, but I can confirm that this can change daily depending on the availability of vaccine in your area.

If you have questions regarding the vaccine for you reach out to your metabolist and potentially primary care physician.

Thanks so much.



Kimberly A Chapman, MD PhD
Geneticist and Metabolist
Medical Advisor HCU America



**Do you or your child have a homocystinuria?
Have you had COVID-19, or the COVID Vaccine?
Tell us about your experience!**

Email us your experience to: info@hcunetworkamerica.org

HCU COMMUNITY COOKBOOK

TRY THESE RECIPES (CLICK THE PICTURE)



JANUARY 4 IS
NATIONAL SPAGHETTI DAY

Carbonara Recipe



JANUARY 24 IS
NATIONAL PEANUT BUTTER DAY

Pea-Not Butter Pie Recipe



Rare Bears for Kids with HCU



We are thrilled to announce that the Rare Bears for HCU Campaign in partnership with Rare Science has been re-opened!

To enroll in the RARE Bear Program and to request a RARE Bear, please click the link and complete the form: <https://www.rarescience.org/hcu/>

- Those who have already received a bear, are not eligible
- Date for gifting will be announced later
- You will **not** receive a confirmation email or be notified when your bear has been shipped

NEWS AND EVENTS

A Sister's Quest for a Cure



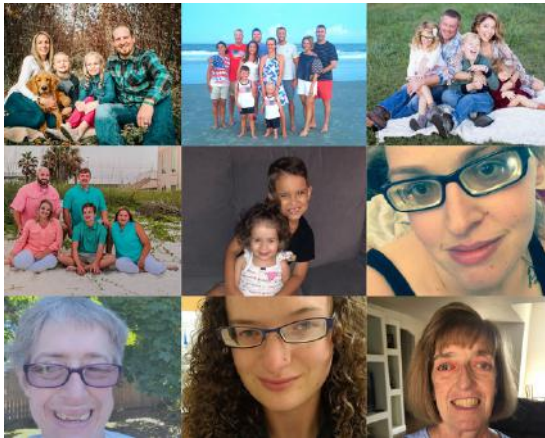
Margie McGlynn, MBA '83 and BS '82, president of HCU (Homocystinuria) Network America, has endowed a professorship in the School of Pharmacy and Pharmaceutical Sciences at University of Buffalo to find holistic solutions for rare genetic diseases like the one that took the lives of two of her sisters in childhood.

Visit <http://www.buffalo.edu/campaign/priorities-and-goals/new-themes-parent-page/helping-people-live-healthier-lives.html> to learn how Margie and the university are working together to provide better solutions for the integrated care of patients with Homocystinuria and other genetic disorders

2020 ANNUAL APPEAL HIGHLIGHTS

Thank you to everyone who donated and helped fund-raise!

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 \$ 4,456 thanks to the 9 families who set up fundraisers on behalf of the organization and to the many others for their individual donations.

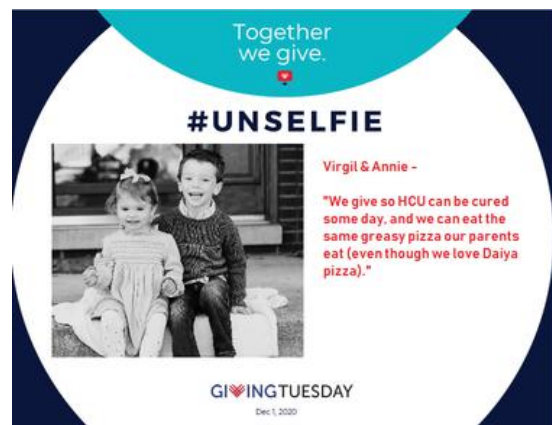


HCU Awareness Month Fundraiser
Participants

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 2020! With these emails we were able to take in \$25,414 to continue our mission in 2020, and we will receive a match for the \$25,000 from our anonymous donors.

We are very thankful for our generous donations from our supporters! This was our third year that we not only met our goal, but surpassed it! We couldn't have had a successful 2020 without you and know this will be true for 2021 as well!

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. The Hawkins' Family also stepped up once again and hosted their second Bears versus Packers Squares Tournament. By the first part of December we were at more than three-fourths of the way to our goal. Thank you to those who set up #GivingTuesday fundraisers. With our great success, it was at this time another anonymous donor came forward and added \$5,000 to our Matching Gift Challenge, making our new goal \$25,000!



Annie and Virgil Hawkins



GI VINGTUESDAY

Giving Tuesday Fundraiser Participants

UPCOMING EVENTS

Come check out our Virtual Homocystinuria Meet-ups!

Join our virtual meet-up for a chance to meet, connect, and learn from other patients and caregivers who are facing similar challenges. Whether it's navigating adherence issues, insurance, clinic visit, or life transitions, you are not alone.

Note our new registration page: <https://hcunetworkamerica.org/virtual-meet-ups/>



HCU Community Virtual Meet-up

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

Sun., January 10th @ 7 pm CST

Mon., January 25th @ 1 pm CST | 7 pm UTC



MMA & Cobalamin Community Virtual Meet-up

Online meet-ups are an opportunity to connect patients and caregivers impacted by methylmalonic acidemia and cobalamin disorders to one another virtually.



Sun., January 24th @ 7 pm CST

HCU Kids Meet-Up

Patients, ages 7 and up with HCU, are invited to a zoom meet-up with familiar and new friends. We will have time to chat and play some fun games!

This meeting is being led by peer leaders, Gabbi Lewis and Sarah May (both who also have HCU).



Mon., January 18th @ 1 pm CST



ADVOCATE FROM HOME!

RARE ACROSS AMERICA

EVERY VOICE, IN EVERY DISTRICT, MATTERS

FEB 22 - MAR 5, 2021

REGISTRATION IS NOW OPEN
[RAREACROSSAMERICA.ORG](https://rareacrossamerica.org)

- ▶ **Make an impact on federal policy from close to home.**
- ▶ **Share your rare disease story.**
- ▶ **Meet other rare disease advocates.**

[Haz clic aquí para obtener información en español y para registrarte.](#)

WHEN

February 22 - March 5, 2021 (Virtual Congressional meetings on March 3rd and 4th)

WHAT

Virtual meetings with your Senators and Representative AND virtual advocacy events. Rare Disease Legislative Advocates (RDLA) organizes meetings for rare disease advocates with their Members of Congress and/or the Member's staff. Meetings will take place virtually on March 3rd and 4th. The RDLA team also helps to prepare advocates for their meetings, provides legislative resource materials, and hosts pre-meeting training webinars.

SCHEDULE OF VIRTUAL EVENTS

Week 1: RARE Readiness

Monday, 2/22: Selfie Monday—Post a picture on social media and tag #RareAcrossAmerica

Wednesday, 2/24: Fast Forward for RARE Practice Your Pitch Webinar

Thursday, 2/25: Virtual Rare Disease Caucus Briefing

Friday, 2/26: Share Your Rare Story—Take a video and post on social media and tag #RareAcrossAmerica

Week 2: RARE Rally

Monday, 3/1: NIH Virtual Event

Tuesday, 3/2: Prepare for Your Virtual Hill Meetings (resources at www.RareAcrossAmerica.org)

Wednesday, 3/3: Virtual Hill Meetings with Senators (12-5 pm ET)

Thursday, 3/4: Virtual Hill Meetings with Representatives (12-5 pm ET)

No prior experience necessary. Registration for all RDLA events are free for rare disease advocates.

SAVE THE DATE: The training webinar will take place on 02/04/21 at 2:00 pm ET, First Time Advocates Webinar on 02/09/21 at 2:00 pm ET, and the Team Leaders Webinar on 02/11/21 at 2 pm ET.



Welcome to The YARR Leadership Academy, a series of on-line Courses offered to a select group of young adults in the rare disease community (ages 18-29). Academy students will learn about the roles and opportunities for patient representation in policy making, drug development and the regulatory process and the steps it takes to get there.

Courses included:

- Welcome and Intro
- Part II: Patient Representativeness, Patient Focused Drug Development, and the Roles for Patient Representatives – March 24th
- Part III: The Basics of Policymaking, Therapy Development and the Regulatory Process – April 7th
- Part IV: Opportunities for Patient Engagement – April 21st
- Part V: Essential Skills and Tools for Career
- Part VI: Capstone Presentation – May 19th Development – May 5th

Examples of Leadership Roles for Academy Graduates:

- Staff or intern for a Member of Congress
- NIH Advisory Council Member
- FDA Patient Representative
- Board of Directors Member for a rare disease patient organization

How to Apply:

- Applications due February 15, 2021
- Submit a letter of reference
- You will need to submit a short essay on what your leadership goals are in the rare disease community.

To learn more and apply, visit:

<https://everylifefoundation.org/young-adult-representatives/yarr-leadership-academy/>

LAND OF THE FREE, HOME OF THE BRAVE

June 26-27, 2021 | Bethesda, Maryland



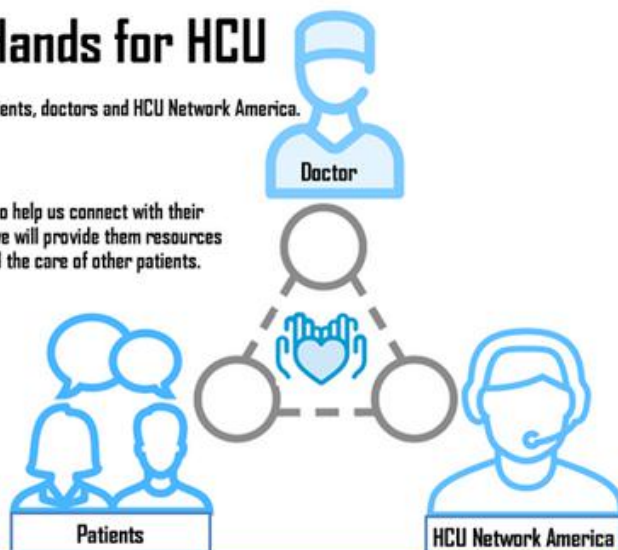
To learn more or register, visit: <https://hcunetworkamerica.org/2021-conference>

GET INVOLVED

Helping Hands for HCU

A collaborative effort of patients, doctors and HCU Network America.

We are asking for patients to help us connect with their metabolic team. In return, we will provide them resources to help better your care and the care of other patients.



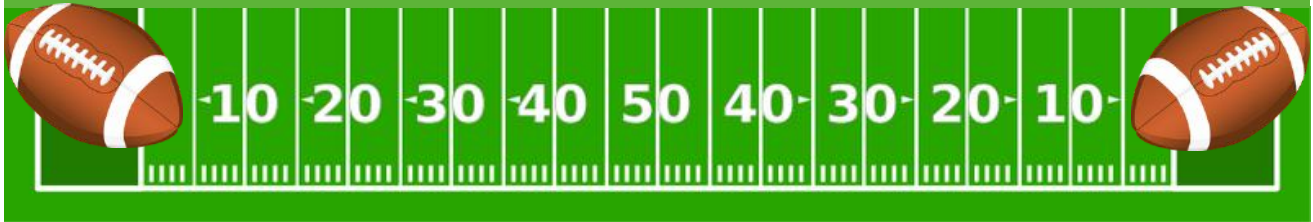
Connecting with clinics is a big priority of ours. Clinic connection allows us to work collaboratively with providers to ensure we are reaching all patients with all the unique needs the Homocystinurias presents.

As a thank you for your participation, **for the month of January, every patient or caregiver that fills out our clinic contact survey will earn a chance to win a \$25 visa gift card.** The winner will be contacted by email the first week of February.

<https://hcunetworkamerica.org/patient-clinic-survey/>

JANUARY FUNDRAISING EVENT: SUPER BOWL SQUARES

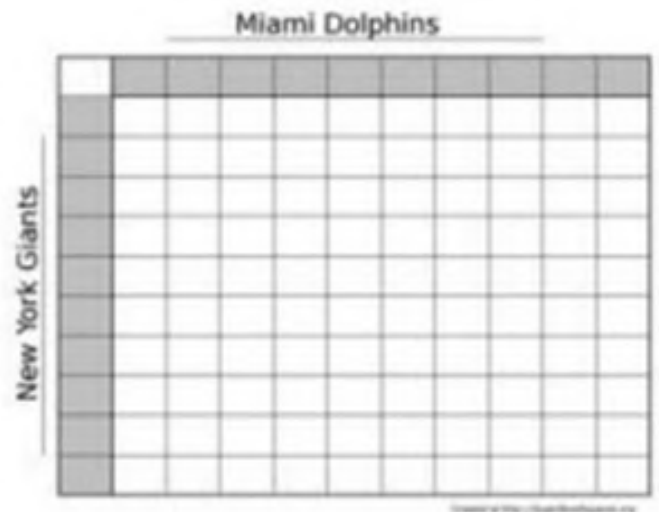
Touch Down!



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 square, 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!



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.

For further instructions and templates:

<https://www.printyourbrackets.com/nflweekly100squares.html>

For online tools, check out:

<http://footballsquaresonline.com>

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



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://hcunetworkamerica.org/contact-register/>

***FOLLOW
US***

