HCU Herald

Presented by HCU Network America

Connecting for a Cure.

There have been a lot of things happening for the HCU community & for HCUNA.

We strive to keep you informed and connected.

Inside this issue:

June's HCU Hero: Joann from Oregon

Heroes of HCU

Joann from Oregon

I was diagnosed with Homocystinuria at age 3, (1974) when I rubbed Trisodium Phosphate (paint thinner) into my eyes. They originally thought it was Marfan's Syndrome, but upon looking at my eyes closer they determined it was Homocystinuria. I was born 10 years after it was discovered in the U.S. and newborn testing was not available at the time. At age 6, my right lens was removed. The surgery was brand new then and so I still have some floaters in that eye. My left lens was removed because of a cataract a few years ago, so I don't have a lens in either eye now, but still see perfectly fine. It is the macula that controls vision, not your lens (which is why when people get Macular Degeneration, they refer to it as tunnel vision.). I see normally with my glasses, but without them if you shine a light in my eyes I see a circle with a whole bunch of dots in the center. I think the darker dots are due to my strokes, but I am not sure about that.

At age 12, I was diagnosed with scoliosis, and they had to do surgery on my upper spine to prevent my lungs from collapsing. Although the rod only goes to my mid-back, my mother still thinks that my whole back was corrected, because the scar goes all the way down my back. If it was corrected completely though I would not be able to move my back at all though, and I can feel where it ends.

At about 19 years old, I had a B-12 problem which caused some balance problems and panic attacks. This is when I was put on Cystadane which helps me to get rid of homocysteine that is trapped in my body. It has a very bitter taste, so I had a hard time finding something to mix it with and finally decided on lemonade. I always liked lemons and they don't require sugar for me. My mother thinks the balance problems and panic attacks was due to a stroke.

I had my first stroke at age 30. I have Grand Mal seizures which trigger strokes, but I haven't had one since 2005. I was 34 at the time of the major stroke which paralyzed my entire left side. Before the major stroke, I didn't believe what my doctors were saying because nobody took the time to look it up and it is so rare. It also helped to have the doctor at OHSU question his students on it. I wasn't taking my medications or following the diet at that time. I was also under a huge amount of stress because of a job I had just left and another I had just started.

I was told as a child if I followed the diet and took the medications then I would never have strokes, and I now believe that. In California they just told me to eat a vegan diet. I didn't have access to the foods that are available now. This is still a touchy subject with my mother, but on a vegan diet even if I go gluten free I haven't been able to keep my levels below 50. Right now with the medical foods and medications my levels are that of someone who has never had the condition. My lowest homocysteine level is 4. You would also never be able to tell I had a stroke by looking at me. Thanks to rehab, and what I can remember from CNA classes that my father recommended, everything is now back.

I now work for The State Oregon Homecare Commission as a Homecare Worker/ Personal Support Worker. I work with those over the age of 65, and those who are disabled on Medicare and Medicaid in their home. I can be trained to do anything that a nurse can do, but most of my employers just want house cleaning and some errands done. They don't realize all that we can do because we are called homecare workers and not caregivers as the title should be. As a Personal Support worker, I work with people who are mentally ill or addicted to drugs. All are my employers are adults. The youngest person I have worked with is 30. My work now keeps me busy enough without outside activities, but I used to run a movie group on Facebook and Meetup that does free screenings before they come out in the theaters. They are still available in many areas globally.

HCU and You: Connecting the Dots

First, I want to say how wonderful it was to meet so many families at the first meeting of HCU Network America. I was struck by how engaged all the families were with the talks, and I overheard so many great conversations where families connected and shared their experiences with one another. The questions that I received highlighted the need for us to understand Homocystinuria at a biological level but also at the level of behavior and social interaction.

Balancing Medically Necessary Behaviors and Everyday Life

Normal, everyday parenting can be exhausting and, at times, feel draining, leaving parents feeling like they need to pick their battles.

It is perfectly normal for children to have disagreements and fights with siblings, to struggle with completing their household responsibilities and chores, and to be angry or meltdown at perfectly reasonable expectations. All kids have responsibilities to contribute to chores at home and to complete homework. On top of the normal stuff, children with chronic health conditions have a lot of restrictions on their behavior like following a strict diet, and they do a lot of things that other kids don't have to do like extra medical appointments and tests.

Parents of children with Homocystinuria often have a question. Many parents have a feeling that their child should have fewer restrictions or fewer responsibilities to "make up" for the extra burden of Homocystinuria. Based on my experiences with families I have a couple of suggestions. I want to recognize amazing effort of families affected by Homocystinuria. I also want to reassure parents that the question of "what is the health condition versus what is normal misbehavior" is present for so many conditions, not just Homocystinuria.

Here are a few general guidelines for families to consider

1) Everyone in a family should contribute to the household including chores. For children, household responsibilities include helping to clean up, respect for the property of others, and
2) Everyone in a household should work towards solving disagreements and arguments without hitting.
3) Children with chronic health problems should help in their daily medical care.

I worked with a family and teenager who was awaiting an organ transplant. While they waited life went on. There were household tasks that needed to be done. The teen was in school and needed to do homework. The struggle for the parents was that death was a possible outcome with or without the transplant. How can you have an argument or let your child be mad at you while death is a possibility? The teenager went without rules and without responsibilities. This went on for several months. The result was family relationships that the teenager and the parents did not like.

I want to encourage families to have high expectations for children. How can families know if their expectations are too low, too high, or about right? First, I want to reassure you that most families of children with Homocystinuria are doing an amazing job. Second, use your existing networks to help guide your level of expectations. For example, talk to the school about the types of behaviors your child can demonstrate. Is the child involved in extracurriculars like sports? Look to see how they solve conflicts or are able to fulfill responsibilities. Third, *go by the numbers*. Measuring behavior is a great way to understand your child's level and to set new expectations. For example, a school IEP might have a goal that says, "Student XYZ current follows instructions to help with classroom clean-up 2 of 10 times during the week, and Student XYZ's goal is to increase participation to 6 of 10 times."

Sincerely,
Ben

Connecting the Dots is an ongoing series of articles that is meant to help answer many of the questions asked by people affected by HCU. Benjamin D. Goodlett, PhD, is a psychology postdoctoral fellow who works with children and families affected by inborn errors of metabolism.

You may watch Ben Goodlett's presentation on HCU and the Brain from our inaugural conference at:
https://www.youtube.com/watch?v=QLo6Ue4hM9s

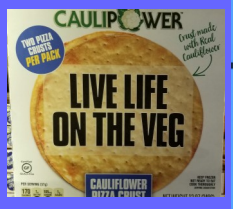
Grocery Store Finds

Grocery List

Date

Non Dairy	Brand	Item	Protein
☐	**So Delicious**	coconut milk yogurt	<1g per 5.3oz cup
☐		coconut milk ice cream	1g per 1/2c most flavors
☐		cocowhip	0g per 2 tbsp
☐		coconut milk beverage, most flavors	<1g per 8oz
☐		Coconut milk ice cream bars, sandwiches	1 g per bar/ sandwich
☐	**Daiya**	cheese shreds	1g per 1/4 cup (28g)
☐		creamy dressings	0.32g per 2 tbsp (30g)
☐		cream cheese all flavors	1.5g per 2 Tbsp
☐		cheese slices	1g per slice
☐		Cheezecake	2.48g per slice (100g)
☐	**Silk**	coconut milk blend	<1g per 8oz
☐	**Rice Dream**	Rice Milk, original	1.4g per 8oz
☐	**Almond Breeze**	almond milk	0.7g per 40z
☐	**Follow Your Heart**	Vegenaise	0.2g per 1 Tbsp
☐		cheese slices	0g per slice
☐		parmesan shreds	0.12g per 28g
☐		cheese shreds	0.2g per 1oz
☐	**Field Roast (Chao)**	Chao cheese	0.05g per slice
☐		Chao cheese, tomato cayenne	0.25g per slice
☐	**Luna & Larrys**	Coconut Bliss Ice Cream, various flavors	1-2g per 1/2 cup
☐			

Breads/ Crusts	Brands	Items	Protein
☐	**Any**	Wonton Wraps	0.75g per wrap
☐	**Food For Life**	GF English Muffins	1g per 1/2 muffin
☐	**Rudis**	GF Bread, raisin	0.89g per Slice
☐		GF Bread, original	0.48g per slice
☐		GF Tortillas	1.01g per tortilla
☐	**Caulipower**	Cauliflower Pizza Crust	2g per 1/3 pizza (57g) Crust Only
☐	**Any**	GF Waffles	1-2g per 2 waffles
☐			



Grocery Store Finds

Grocery List

Date

Frozen Vegeta-	Brands	Items	Protein
☐	**Dr. Praeger's**	Carrot Puffs	1.75g per 13 pieces
☐		Potato Littles	1.2g per 2 pieces
☐		Sweet Potato Littles	1g per 2 pieces
☐		Hashbrowns	2g per 3 browns
☐		Kale Puffs	2g per 14 pieces
☐	**Alexia**	Mashed Cauliflower	1g per 1/2cup (113g)
☐		Mashed Butternut Squash	<1g per 1/2 cup (113g)
☐		Riced Vegetable Pilaf	1g per 1/2cup (113g)
☐	**Green Giant**	Riced Cauliflower, original	2g per 1 cup
☐		Cauliflower Tots	2g in 6 pieces
☐		Zucchini Spirals	1g in 3/4 cup
☐		Butternut Squash Spirals	1g in 3/4 cup
☐		Carrot Spirals	1g in 1 cup
☐	**Birds Eye**	Mashed Cauliflower	2g per 1/2 cup
☐		Mashed Cauliflower, sour cream & chives	3g per 1/2 cup
☐		Mashed Sweet Potatoes and Carrots	1g per 1/2 cup
☐		Riced Cauliflower, original	2g per 3/4 cup
☐		Riced Cauliflower, Italian Cheese	2g per 3/4 cup
☐		Riced Cauliflower, Roasted Garlic or Herb	1g per 1 cup (123g)
☐		Flavor Full Buffalo Cauliflow-er	1g per 1 cup (94g)
☐		Flavor Full Steakhouse Green Beans	1g per 3/4 cup (83g)
☐		Flavor Full Ranch Cauliflower	2g per 1 cup (94g)

Potatoes	Brands	Items	Protein
☐	**Alexia**	Potato Puffs, Sweet Potato	1g per 2/3 cup
☐		Potato Puffs, Seasoned	1g per 14 pieces
☐		Fries, Yukon Select	2g per 3oz (24 pieces)
☐	**Ians**	GF Alphatots	2g per per 14 pieces
☐		GF Onion Rings	1g per 5 pieces

Grocery Store Finds

Grocery List

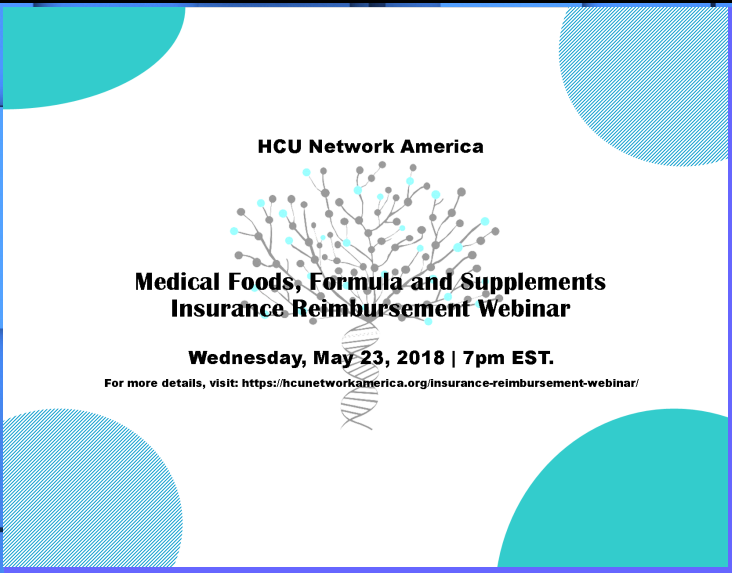
Date

☐	**Any**	Shoestring Fries	1g per 45 pieces
☐	**Ore-Ida**	Mini Tator Tots	1g per 87g (19 pieces)
☐		Sweet Potato Fries	1g per 3oz (22 pieces)
☐		Hashbrowns, Diced	1g per 2/3 cup (85g)
☐		Hashbrowns, Shredded	1g per 1 1/4 cup (85g)
☐		French Fries, Country Style French Fries	2g per 84g

Meatless	Brands	Items	Protein
☐	**Morning Star**	Bacon Strips	2g per 2 pieces
☐	**Dr. Praegers**	Mushroom Risotto Burger	3g per burger
☐		California Veggie Burger	2g per burger
☐			

Special Finds	Brands	Items	Protein
☐	**Konjac**	Shirataki Noodles	0g per 83g (1/3 package)
☐	**Dove**	Chocolate Covered Mango Sorbet Bars	1g per bar
☐			

In case You Missed It!



Wednesday, May 23 Raenette Franco gave a very informative webinar on Medical Food, Formula and Supplements Insurance Reimbursement for the Homocystinuria community. During the webinar she walked us through insurance terminology, the differences between medical and pharmacy coverage, reimbursement issues between insurance company and supplier, billing discrepancies, verifying insurance benefits before placing an order and medical food exclusion removal. At the end of the webinar, she opened the floor to anyone who had a question about the information she spoke about, or their own personal experiences.

Were you unable to view the webinar? You can watch our recording here: https://youtu.be/JC_Z5S1VkvI

HCU Network America's Partnership with Raenette Franco

We realize that food and formula coverage varies greatly by state. For this reason, HCU Network America has contracted Raenette Franco of Compassion Works Medical LLC to help assist you in getting low protein foods, formula and supplements covered. She will also work with you to find a policy that meets your individual needs if you don't currently have one, or are struggling to find one.

Who is Raenette Franco?

A native New Yorker, Raenette Franco came to New Jersey in 2001 and landed a career as Medical Biller Insurance Specialist/Consultant. Her background consists of hospice and palliative care, bariatric surgery, and other medical specialties geared around insurance coverage. Raenette landed a job within the medical food arena in 2011 as a medical food insurance specialist that captured her heart. Inspired by helping others, Raenette expanded her career as founder of Compassion Works Medical. Raenette received her Certification as a Certified Biller Coder Specialist (CBCS), from the NCCA Accreditation, NJ. Her big heart and deep passion is dedicated to helping patients, dietitians, and physicians avoid the difficult tasks of insurance coverage and reimbursement issues for medical foods/enteral nutrition. Raenette's specialty is working directly with Medical Food coverage for both formula and low protein foods.

Mission:

Compassion Works Medical has a special mission to change the lives of the rare genetic disease community by supporting a genuine helping hand with insurance coverage challenges. Over the past years at Compassion Works Medical, a blessing in disguise had come across Raenette's path – she has expanded her support to all kinds of special diseases that require medical foods and enteral nutrition, including cancer, ALS, TBI, etc. It is truly rewarding and her inspiration grows stronger and stronger to help those people find a way to afford and stay on diet.

"We don't take NO for an answer along with compassion that makes all of us successful"!

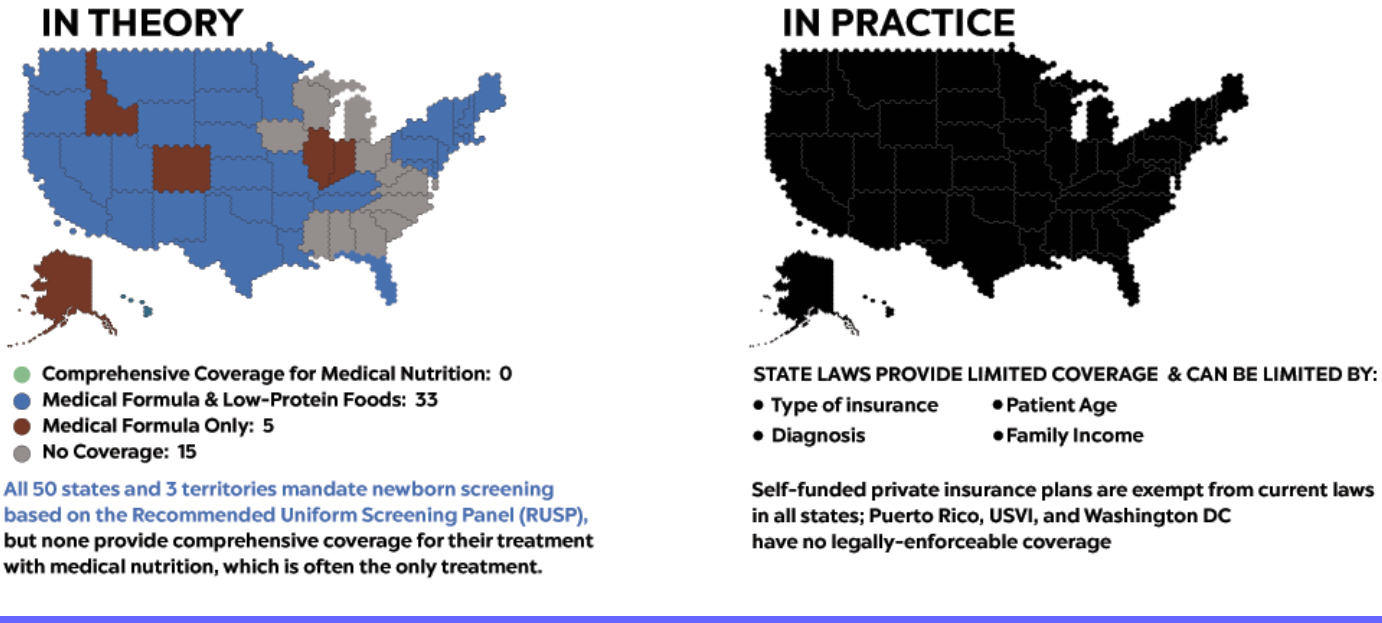
How to get in touch with Raenette:

raenettef@compassionworksmrs.com
(973) 832-4736

Ways to Get Involved

PATIENTS & PROVIDERS FOR MEDICAL NUTRITION EQUITY

The **Patients and Providers for Medical Nutrition Equity Coalition** is a group of patient organizations (including HCU Network America) and clinicians involved with disorders that require medical nutrition to maintain health. We are working to pass the **Medical Nutrition Equity Act** (S-1194, HB-2587), which will extend the coverage for medical nutrition that Congress passed in 2016 for TRICARE patients to other Insured Americans who have inherited metabolic disorders/inborn errors of metabolism and other disorders on the Recommended Uniform Screening Panel, medical and surgical conditions of malabsorption, Immunoglobulin E and non-Im-mu-no-glob-u-lin E-mediated allergies to food proteins, inflammatory or immune mediated conditions of the alimentary tract, and other diseases or conditions determined appropriate by the Secretary of Health and Human Services.



For many in congress they know very little, if anything about Homocystinuria or other inborn errors of the metabolism. Your stories of inadequate medical food and formula coverage will help paint the picture of the struggle we face daily feeding ourselves and our children with these rare and life threatening conditions.

Please share your stories about how you have been impacted by inadequate medical food and formula coverage! Visit https://medicalnutritionequityfor.us/share-your-story/ and share your story now

To learn more about Patients and Providers for Medical Nutrition Equity Coalition and how you can get involved, please visit: Https://medicalnutritionequityforus.com

Newborn Screening Survey for Classical Homocystinuria

But we have newborn screening for HCU...

According to recent statistics, approximately 25-50% of patients are missed by newborn screening for Homocystinuria. There are multiple factors that can play into these numbers. Currently it is federal mandate that all states screen for Homocystinuria through the newborn screening test, but there are no set standards. Meaning, every state or region can set their own methionine cut offs. A handful of states also do tier two testing—meaning they have a second round of newborn screening, making it more likely for homocystinuria to be picked up. Another factor that plays into the effectiveness of the test, is how elevated the patients levels are at the time of the test. Patients who are pyridoxine (B6) responsive, or have more functioning CBS enzyme, are less likely to be picked up by the newborn screening

So how can you help?

Talk to your geneticist about the newborn screening survey and urge them to complete it! This will help us build support for changes to the process to increase the likelihood that HCU patients will be diagnosed at birth.

Here is the letter portion we would ask you to give to your clinic, followed by the survey form:

The Letter:

To Whom this may concern,

I would appreciate your support in answering a brief survey to help support efforts to improve newborn screening for classical homocystinuria.

I have been working with HCU Network America, a patient advocacy and support group for Homocystinuria (HCU), for whom I serve as a medical advisor. One of their key goals is to improve newborn screening for HCU, as it is estimated that over half of patients are missed by the current screening process and often are not diagnosed until they have developed serious clinical symptoms. To build support for an improved process, we are collecting information on patients missed by the current screening process, which we intend to then publish in a consolidated case report.

Could you please support our efforts by completing the attached brief questionnaire, and sending it to me via e-mail at:

FICICIOGLU@email.chop.edu

Sincerely,

Can Ficicioglu, M.D., Ph. D.
Director of Newborn Metabolic Screening Program, Children's Hospital of Philadelphia

Newborn Screening Survey for Classical Homocystinuria

Survey on Classical Homocystinuria (HCU) Patients Missed by Newborn Screening

Do you have any patients with classical HCU missed by NBS and diagnosed later based on symptoms?

( ) Yes ( ) No

If yes, at what age were the patients diagnosed, and what year were they born and in what state?

Age at diagnosis (mos.) ____ Year of birth _____ State born_____

Age at diagnosis (mos.) ____ Year of birth _____ State born_____

Age at diagnosis (mos.) ____ Year of birth _____ State born_____

Age at diagnosis (mos.) ____ Year of birth _____ State born_____

Age at diagnosis (mos.) ____ Year of birth _____ State born_____

Would you be willing to provide information to contribute to a "Case Report" we plan to publish on patients missed by Newborn Screening?

What is the name and address of your clinic and the best contact person for further information:

Clinic Name _____

Clinic address _____

Contact Person:

Name _____

E-mail _____

Phone _____

Please send completed survey to Dr. Can Ficicioglu at **ficicioglu@email.chop.edu**

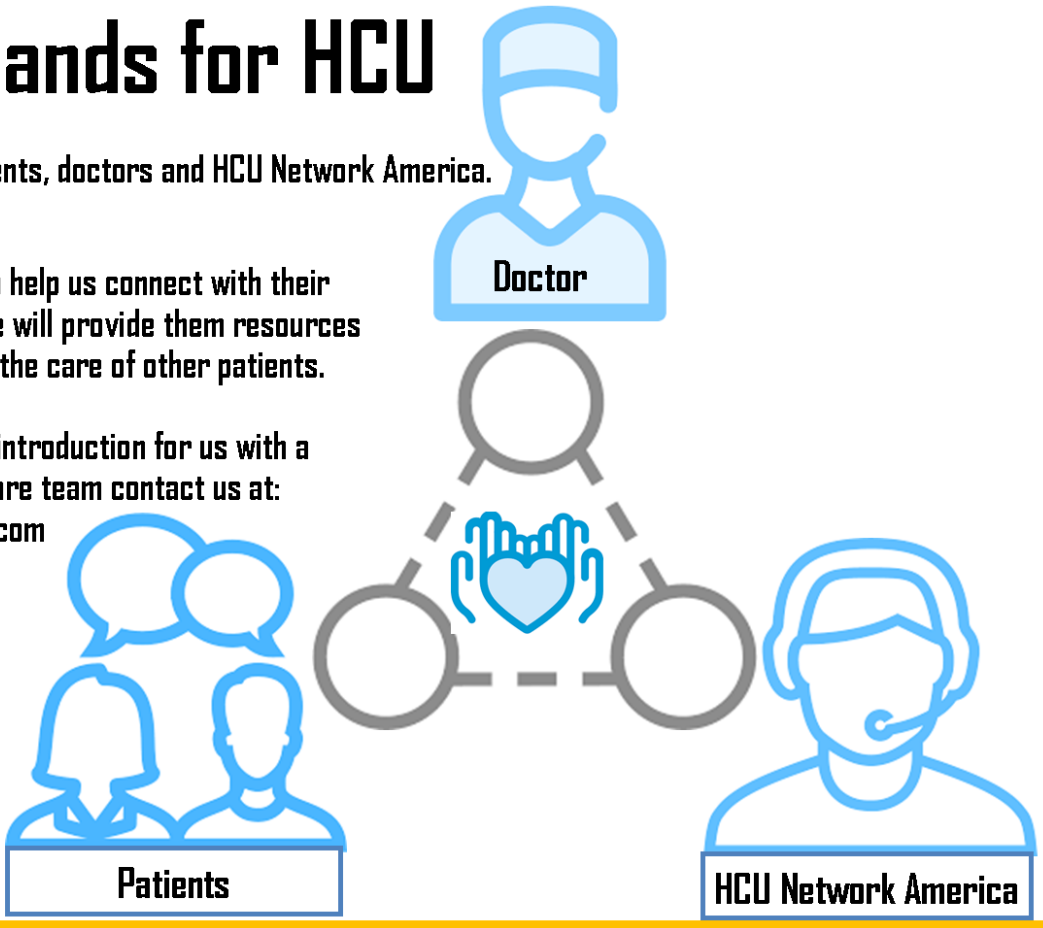
Ways to Get Involved

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.

If you are willing to make an introduction for us with a member of your metabolic care team contact us at: HCUNetworkAmerica@gmail.com



Natural History Study

Current sites include: Boston, Philadelphia & Atlanta.

Joining the Natural History Study allows researchers to find out more about Homocystinuria and issues that patients face. Natural history studies help drive new therapies and a cure! If you qualify, we highly suggest you participate if there is a center in your area. You do not have to be a patient at one of these clinics to participate.

Contact Register

Did you know that just under one year ago we launched our 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 to. By registering, you will be also be able to identify other affected patients in your state and request their contact information, and you will 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 and provide resources for patients and families, create connections, and support advancement of diagnosis and treatment of HCU and related disorders. In order to succeed in our mission, the input you provide helps us plan events, develop resources and educational tools, and to help to ensure everything is being done to guarantee 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/

Coming Soon: HC&U Podcast

Current research

Interviews

Low protein Recipes

Day to day life

And many other topics!

COMING SOON!

What is HC&U?

HC&U is a podcast about Homocystinuria, sponsored by HCU Network America and hosted by Ben and Lindsey Massengale. Ben was diagnosed with HCU at birth, and Lindsey had no idea what HCU was. Then Lindsey met Ben, they got married, and Lindsey had to learn how to cook vegetables in a bajillion different ways. This podcast will include interviews with different professionals in the field, a low protein recipe each episode, and even personal stories from Ben's experience with HCU. We are open to suggestions on topics you would like to hear, so please email us any ideas you have. Watch our social media sites for the podcast release date!

HC&U

Twitter: @hcupodcast

Facebook:
HC&U: A Homocystinuria Podcast

hcupodcast@gmail.com

Meet Nick Lunardini:
Interim Coverage

We can officially say that Danae' Barkte, our executive director is quickly approaching her due date! With that being said, we would like to introduce you to Nick Lunardini. Nick will be covering the day-to-day operations that Danae' typically preforms while she is on her maternity leave.

About Nick:
My name is Nick Lunardini and I am honored to be part of the HCU Network of America, if even for a brief time. I am the father of three young children and a small business owner. I believe in giving back to the community and building relationships with others, no matter the context.

My experience includes over 15 years of personnel management experience, as well as product management, purchasing, and customer service. My previous positions have allowed me the opportunity to live in many areas of the country and be part of many new experiences. I look forward to being a part of the HCU Network community.

Amazon Smile

Shop Smile.Amazon.com when purchasing through Amazon and 0.5% will be donated to HCU Network America when you designate us as your charity of choice.

Click here to find out how!

Company Matching

Did you know that many companies big and small may match donations made to HCU Network America?

Click here to find out more

Click to donate directly

We'd like to thank the following content contributors:

Editor in Chief: Danae' Bartke
Heroes of HCU: Joann from Oregon
HCU and You: *Connecting the Dots*: Ben Goodlett
Grocery Store Finds: Amber Gibson
HCU Podcast: Ben Massengale