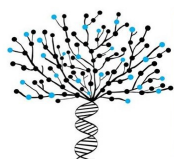


HCU Herald

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:

HCU Hero: Samantha

HCU and You: Recipes from the Kitchen

HC&U: A Podcast About Homocystinuria

Back to School With Amazon

Chicago Marathon Challenge

HCU Awareness Month

Save the Date: Rome 2019

Ways to Get involved

Contact Register



Heroes of HCU

Samantha From Kentucky



I began my last semester of college excited for the year ahead, doing normal things for a college senior, thinking about life after graduation and hanging with my friends. I was looking forward to my last performance of Step Sing, a campus - wide production unique to Samford University that engages with the community and raises funds for diverse organizations and is embraced by the student life and philanthropic missions. I was also looking forward to working

with the athletic department like I had for the previous three years. It was during the three weeks of preparation for Step Sing, which includes practices that lead up to a performance, that many of my friends were getting sick. I hadn't been feeling well for some time, so I thought I just may have caught something like my friends had, i.e. some kind of virus. In this case though, I thought my usual sinus headaches were a little worse than and more frequent than I had experienced in the past. I was experiencing migraines that would go down my body and I was constantly feeling nauseous with these headaches. Until that time, I had not experienced the nausea associated with headaches. They came more frequently and eventually disrupted my lifestyle as they continued throughout the remainder of the semester, occurring at least once every month if not more. I went to the health clinic on campus and they kept telling me there was nothing wrong with me. Unfortunately, with my previous healthcare concerns, they referred me to my previous diagnosis, Chiari, and assumed they could not treat me without reviewing the symptoms of that disease. As one that has experienced everything I have, I am familiar with the issues with clinical diagnosis of rare diseases, and my unique medical concerns.

Luckily my mom liked to visit me every two weeks and she felt I also had an almost jaundiced look and weakness she hadn't seen in years. So as usual my mom and I decided it would be best to schedule a follow up with my neurosurgeon in Iowa after graduation. I have an OC3 fusion to correct problems due to an abnormality in the cranial cervical junction also associated with Chiari Malformation, and with that follow up in May he told me my fusion was still intact and on his end everything was good. But Dr. Menezes treats so many of us with rare genetic conditions that he felt a visit to the pediatrics specialty group genetics clinic to get follow up blood test from when I was 13 years-old was necessary to confirm the source of my issues. He felt that after my initial surgery in 2006 I may have Loey-Dietz but at the time there wasn't a test available to confirm the diagnosis. After that blood test came back I was diagnosed with Loeys-Dietz Syndrome (LDS), a connective tissue disorder similar to Marfan's, and Homocystinuria. I didn't know what either of those things were, but my mom and I were told the best place for us to go was to John Hopkins University Hospital in Baltimore, Maryland where Dr. Dietz was. What a blessing! Once Dr. Dietz and his assistant, Gretchen, heard about my case and with no other alternative locally with LDS and HCU they also set us up with Dr. Gunay and a nutritionist, Celide Koerner, who are both very well educated with HCU. Having everyone on the same page (team) was incredibly educational and convenient. They explained to me that having both LDS and HCU is very rare, but for me that's normal because everything I have I feel like has been unique to me, as my mom and I have called it part of the "Samantha Syndrome". Dr. Dietz, and others, refers to me as their unicorn.

Since this diagnosis came to me as a huge surprise, it's been so hard to explain to family and those closest to me. Having HCU has made a huge impact on my life mainly because I was not diagnosed with it until I was 23 yrs. old, and even though I really had no symptoms of the disease that I noticed, I had to totally change my diet. Before being diagnosed, almost every day for lunch I had a turkey and cheese sandwich, which obviously is no longer my regular lunch. Changing my diet hasn't been the easiest thing especially with my parents owning a Mexican fast casual restaurant, Salsarita's Fresh Mexican Grill.

Heroes of HCU

Samantha From Kentucky - Continued

I have found a way to continue to eat there, thanks in part to nutritional calculators found on Salsarita's website. So before I decide what to eat I input what I'm thinking of getting and that's how I decide what to eat from there. My family has helped me make the transition really easy. My parents and grandparents are always helping me find new recipes to try, which for me has been a good experience because having to try different things to see if I would like them is not something I would have done without this diagnosis. Also I am "no fan" of those supplement drinks and honestly just the look grosses me out, so I have found ways of still taking them but without having to look at or taste them, I make my own smoothies with them, which taste and look a lot better. Thanks Mom for the counseling. My Mom even goes to Whole Foods to find energy shots and such, so I know I'm not alone when she gags them down with me. Sometimes weirdness is awesome.

I have actually over three years dropped my homocysteine count from 250 to below 50, with the help of folic acid, B6, B12 shots monthly, betaine, and the change of my diet. The hardest part for me has been the change of my diet, as a 25 year-old and having to change everything from what I used to eat. Seeing your friends enjoy things you used to be able to isn't the easiest but I have such a great support system that makes everything easier.

I have been so thankful for my doctors and nutritionist at John Hopkins who have connected with me some girls in the HCU Community, because for me it's easier to hear stories about other people getting through this knowing that I am not the only one having to deal with this change in diet. And the time spent educating us has been invaluable and meant the world to me and my family. I really have the best medical support system in the world and I am thankful. The real struggle for me has been cutting back my protein intake because I have found out it's so much harder than it looks. Since my diagnosis I have been reading more and more labels on food and going to restaurant websites trying to find their nutrition information so my family can still get things they want to eat and I can get what I can have as well. I have also learned from guidance about having a nutritionist you can trust and include in your wellness approach is beneficial for anyone with such a change in lifestyle. I'm in this for the long run with a positive and healthy outlook. It's so important to seek out and become educated. How can I be successful in this journey? Be resourceful, be confident and lean on friends, family and others living with HCU. Of course be honest with yourself and your medical team and lean on them and keep an open mind.

That's my recipe for living well!

HC&U: A Podcast About Homocystinuria



First Episode Released July 15th!

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!

Current research

Interviews

Low protein Recipes

Day to day life

And many other topics!

HC&U

Twitter: @hcupodcast

Facebook:
HC&U: A Homocystinuria
Podcast

hcupodcast@gmail.com



HCU and You: Recipes from the Kitchen



Roasted Farmers Harvest Pasta

This recipe is a great way to use vegetables from your local farmers or farmers market. You can use any of your favorite vegetables and it will be delicious! You may need to adjust the overall protein or PHE amounts depending on the vegetables used, but it should not be a significant difference. It can be made as a hot pasta dish for a nice dinner for two or can be served cold as a pasta salad for the cookout.

Ingredients

68g sliced bell peppers
125g sliced zucchini or yellow squash
50g sliced onion
2 cloves unpeeled garlic
65g sliced button mushrooms
80g cherry tomatoes
3 tbsp. olive oil
2 tbsp. Sherry cooking wine (optional)
Salt and pepper to taste
1 tsp. chopped fresh rosemary
2 tsp. chopped fresh basil
2 tsp. chopped parsley
2 tbsp. balsamic vinegar
Pasta of your choosing

1. Preheat oven to 375o Fahrenheit. Line a baking sheet with foil and set aside.
2. In a medium bowl combine the peppers, squash, onion, garlic cloves, mushrooms, and cherry tomatoes. Add the olive oil and sherry wine, if using, to the vegetables. Toss to coat the vegetables evenly. Add salt and pepper to taste and toss again to mix. Remove the mushrooms and tomatoes and place in a small bowl and set aside. These will roast quicker than the rest of the vegetables, so we will add them later.
3. Place seasoned vegetables, except the mushrooms and tomatoes, in prepared baking sheet, making sure the garlic is placed in the center of the pan. Place in oven and roast for 10 minutes.
4. Remove pan from oven and add the mushrooms and tomatoes. Lightly mix the vegetables. Be careful not to burn yourself on the hot pan! Place pan back in oven and roast another 10 minutes.
5. Remove pan from oven and add the chopped rosemary, basil, and parsley. Lightly toss.
6. Once the garlic cloves are cool enough to handle, remove the skin of the cloves. The garlic should now be nice and soft. Chop the roasted garlic and gently toss with the other vegetables. You can now add the roasted vegetables to your pasta. Add a little more olive oil to the pasta and a splash of balsamic vinegar. Serve warm with nondairy cheese and low protein breadsticks.
7. If you want to make this a pasta salad, allow the vegetables to cool completely then refrigerate. Add to your cooled pasta right before serving. You can also use the pasta salad dressing of your choosing.

127g roasted vegetables = one serving

2.03g protein per serving

Back To School With Amazon



**Shop at AmazonSmile
and Amazon will make a donation to HCU Network America!**

Do you shop on Amazon? If so, you can have a portion of your eligible purchase price come to HCU Network America! Instead of logging into the normal Amazon website, simply type this link into your browser for the portal to the AmazonSmile for HCU Network America.

<https://smile.amazon.com/ch/81-3646006>

Amazon will donate 0.5% of the price of your eligible AmazonSmile purchase to **HCU Network America** whenever you shop AmazonSmile. AmazonSmile is the same Amazon you know. Same products, same prices, same Amazon Prime benefits – but it helps us!

After your initial visit to the link above, you can go directly to [Smile.Amazon.com](https://smile.amazon.com) (make sure you bookmark this site) and your continued shopping will benefit us.

Please visit and encourage others, too!

Kristen's Chicago Marathon Challenge



Kristin is the Vice President of HCU Network America, an HCU patient, and an avid runner. On October 7th, she'll be running in the 2018 Chicago Marathon.

How did she get into running?

Kristin got into running about 12 years ago. She says her love of running started with the Broad Street 10 miler in Philadelphia. The thing she remembers most about that race was the expression of pride on her parent's faces when she crossed the finish line. She later found running could be an outlet for dealing with tough times, such as the loss of her father in 2007 to cancer or the daily struggle with managing the HCU diet.

Inspired by the 1st HCU Conference in 2011, she started thinking of ways she could connect her passion for running with her desire to help others struggling with rare disease. While doing some research on the internet, she discovered the Running for Rare Disease (RFRD) Team, a group of dedicated Individuals who run marathons to raise awareness and funding for the rare disease community. In 2012, she joined the team first as a patient partner. Later that same year, she was asked if she'd like to join the team as a runner in the 2012 Boston Marathon.

Fast forward to 2018, Kristin has now completed 12 full marathons and has raised over \$25,000 for the National Organization of Rare Disorders (NORD) and \$5,000 for the American Liver Foundation. The funds she's raised for NORD have contributed to a restricted research grant for HCU and have helped support NIH's undiagnosed diseases program. She's also completed multiple half marathons, several century bike rides, and even a triathlon.



What are her hopes for the race?

This fall Kristin hopes to cross the finish line at lucky #13, the Chicago Marathon. This will be the first time in 2 years that Kristin has set out to complete a full marathon distance, which is 26.2 miles. Since she's just getting back into running from a bit of a break, the only running goal she's set for race day is to reach the finish line before the course time limit of 6 hours 30 minutes.

What are her fundraising goals for this Chicago Marathon Challenge?

Kristin is setting an aggressive fundraising goal of \$10,000 for HCU Network America. She will be dedicating each mile of the race to a different HCU patient. More information on how you can get involved as a patient partner and a link to her fundraising page will be coming soon! Watch for updates on the HCU Network America Facebook page and in the next HCU Herald.

HCU Awareness & You

GO BLUE FOR HCU

October is HCU Awareness Month

HCU Network America — Hcunetworkamerica.org — Hcunetworkamerica@gmail.com

**Start planning your Homocystinuria Fundraiser now!
Have a fundraiser planned? Email us and we will help spread the word.**

Need an idea?

<https://hcunetworkamerica.org/fundraising/>

Art Show/Sale

Backyard Carnival

Walk/Run, 5K or Marathons

Restaurant Night

Etc.....



Need help? Email us - we will guide you through the process.

2019 Rome Conference



Save the Date: Patient - Expert Meeting 2019

HCU Network Australia will co-host the 3rd International Homocystinurias Patient-Expert Meeting in Rome, Italy, on Thursday 28th February – Friday 1st March 2019. The meeting will bring together leaders in the field of homocystinuria research, clinical care, nutrition and advocacy for a two day conference. The meeting will be held in conjunction with Rare Disease Day 2019 on 28th February.

The following sessions are planned for the Patient-Expert Meeting 2019.

- Session 1: State of the Art Lectures
- Session 2: New Developments
- Session 3: Newborn Screening
- Session 4: Long-term Outcomes
- Session 5: Disease Causing Mechanisms
- Session 6: Voice of the Patients
- Session 7: Dietary Intervention
- Session 8: Novel Treatment Developments
- Session 7: Interactive Panel Discussion

See the link below for more info:

<https://www.hcunetworkaustralia.org.au/patient-expert-meeting-2019/>

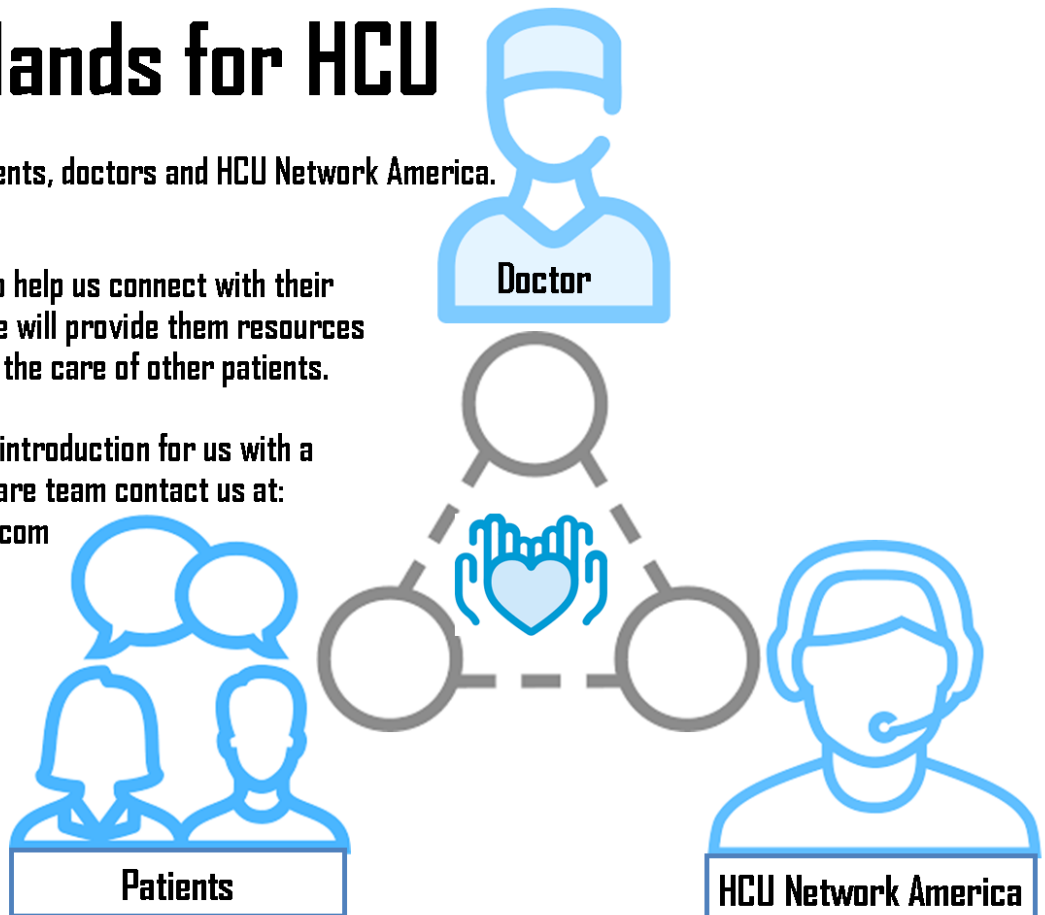
Ways to Get Involved

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



HCU Network Australia Patient & Caregiver Survey

experience of
diagnosis
and
patient
satisfaction.



PATIENT & CAREGIVER SURVEY

There is currently limited evidence showing the considerable length of time taken in some cases to reach a diagnosis of homocystinuria and the level of patient satisfaction regarding access to information, treatment options and medical care. This survey is important to highlight the current situation and has been prepared in consultation with a Key Opinion Leader (KOL) in Europe together with input from HCU Network America, to ensure the information gathered can be used globally to help support improved care for all HCU patients.

We ask no matter what your experience, good or bad, you complete the survey.

To complete the survey click [here](#).

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. 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/>

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: Maeve from Conneticut

HCU and You: *Ask Methia*: Angela Pipetone