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:

Heroes of HCU

Ben from Massachusetts

Hello, my name is Ben, and I have lived with classical homocystinuria (HCU) ever since my diagnosis at birth almost 27 years ago. I was fortunate enough to have been born in Massachusetts, one of the states that tested for HCU at the time as part of the newborn screening program. As the firstborn in my family, and the first member of my family's ancestry known to have HCU, it was certainly shocking, confusing, and nerve-wracking for my parents to discover I had inherited a genetic disorder that could result in dangerous blood clots, skeletal complications, issues with my eyes, and other serious conditions. Fortunately, we were referred to Dr. Mary Ampola, a "world famous specialist" at Floating Hospital for Children at Tufts-New England Medical Center. Dr. Ampola educated my parents, and later, me, on HCU, giving us confidence that I would be able to live a normal life if I followed a low protein diet and took the proper medications and supplements. She provided support and care that was truly exceptional, and I have met very few other people who have exhibited the same genuine care and compassion that Dr. Ampola did for all her patients. Without her, my parents would have been lost after the diagnosis and I would not be as healthy as I am today having had her guidance and expertise to ensure a complication-free life thus far with HCU. Dr. Ampola may have retired in 2004, but her involvement throughout my early life continues to have an impact on my health and management of HCU to this day.

Because of people like Dr. Ampola and my family members, I have led a life with little direct impact felt as a result of HCU. However, that is not to say it has not been tough at times. While I have suffered no known physical impacts resulting from the disorder, I have certainly gone through periods that have been tough psychologically. As a kid, it was sometimes difficult to go to school or a friend's party and have to eat different foods from everyone else. Growing up, it was not as common for children my age to have specialized diets, either by choice or as prescribed because of a medical condition. As a result, I was often the lone kid foregoing ice cream at a birthday party or scraping the cheese off my pizza in the lunch room. Other times, it has been hard to find foods I could eat when at a restaurant or looking for new meals to make at home. However, I have, for the most part, come to embrace my disorder. Specialized diets have become more common, which means others are more accepting and there are more menu options available when I go out to eat or shop for groceries.

Outside of my own experience with HCU, I have been impacted by its effect on my youngest sister, Gabbi, who also has the disorder, having been diagnosed pre-birth by amniocentesis. Gabbi, who is now 16, has, unfortunately, gone through a lot, including extensive surgery on her back to install titanium rods and screws to correct severe scoliosis. Her courage and motivation throughout the recovery period following the procedure has been inspiring and, while I always try to be a role model for her, I will be the first to admit sometimes she's been the one pushing me to be better about how I handle HCU.

Heroes of HCU, cont.

When thinking about my successes as someone living with HCU, I think the key is that I do not typically think of them as being connected to my disorder – I treat achievements as anyone would. In doing so, I hope it shows others, especially parents of children with HCU, and the children themselves, that your life does not have to be lived out in spite of the disorder or, in other words, HCU is not something that should be viewed as an inhibitor or limitation. I do, however, think living with HCU has ultimately made me a more resilient and responsible person, as it has added a degree of adversity to my life. But I see this as a positive. That resiliency and responsibility has allowed me to graduate with high honors from Rensselaer Polytechnic Institute, climb all 48 of the mountains making up the 4000 Footers of New Hampshire, and pursue a Master of Business Administration at Babson College's F.W. Olin Graduate School of Business. In fact, while I am participating in a natural history study and soon a clinical trial to help find a treatment that would all but eliminate the risks associated with unmanaged HCU, I am not sure I would go back and change my past to forego having it in the first place. It may sound odd, but while I hope to help others avoid the disorder and its ill effects, I think having HCU has helped shape me into who I am today and that is something I would not change.

Overall, I think my story has themes that lead to much of the advice I would provide patients and parents who have HCU as part of their lives. First, take advantage of your support network and the robust community that has formed around this disorder. Knowledge is power, and the more you can learn about managing, treating, and living with HCU, the better off you or your loved one will be throughout life. Second, while HCU can easily be seen as a weakness, turn it into a strength. Sometimes, living with the disorder can be tough. Instead of being discouraged or depressed by those situations, try to see them as opportunities to build resiliency and responsibility. And third, if willing and able, find ways to use your perspective to help others. For me, that means participating in the ongoing trial to hopefully find a cure that allows others with HCU to live a life free of highly restricted diets and the risk of serious complications. It also means I am happy to be a resource or connection for anyone in the HCU community, patient or parent. Please feel free to get in touch on Facebook/Messenger (you will find me as Benjamin Lewis). Thank you and hope this is helpful to you on your journey with homocystinuria!

HCU and You: *Connecting the Dots*

Comfort through Connecting

What's Homocystinuria? Homocystinuria is a rare autosomal recessive disorder of methionine and homocysteine metabolism. It affects the eye, skeleton, nervous system, and vascular system. Discussion of Homocystinuria often focuses on the biochemical and medical consequences of accumulation of various metabolites.

Missing from the discussion is the social and emotional effect on children and parents. Any diagnosis or health problem can feel isolating. Social isolation - it can occur alone or surrounded by friends and family. Chronically feeling lonely can lead to dark predictions about the future, feelings of inadequacy, and shame. For parents of children with rare diseases, the isolation can be that much more real because their community, family, and likely even their doctors have not experienced it before. Over and over, people with Homocystinuria must be the ambassadors of their diagnosis, explaining a condition that their friends, school, and work colleagues have never heard of before.

Acceptance of feelings. Some families find comfort in knowing that they are not the only ones experiencing feelings of isolation because of Homocystinuria. More research on the experience of isolation in families effected by Homocystinuria is needed, but times that families might feel particularly lonely are the weeks after the initial diagnosis, at major transitions such as starting school, and planning for the future and how Homocystinuria might impact things like having children. It can be easy to fall in to a "mind over mood trap" where you try to will yourself into no longer feeling isolated. If you are a parent, it's inevitable that you will feel isolated regardless of rare disease or not.

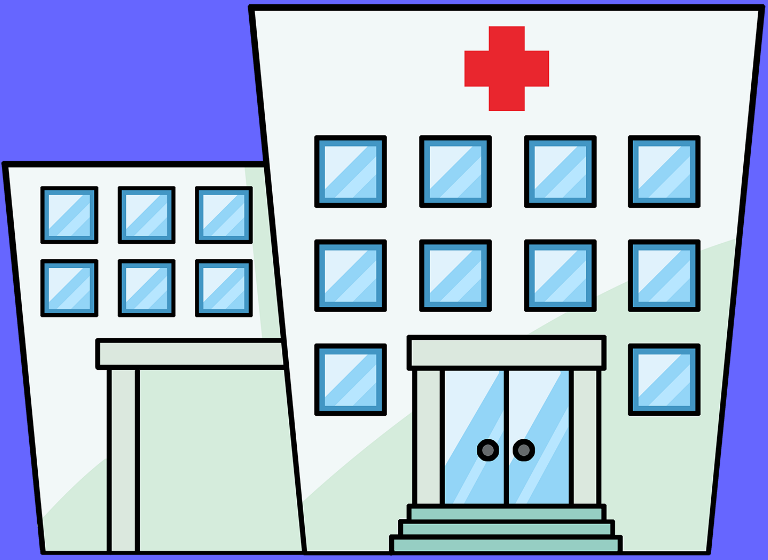
Redefine the meaning of community. Finding other people and other families with the same rare disease can be hard. HCU Network America is working to be the patient support group for people and families affected by Homocystinuria. You may have heard the term "allied disorder" used to describe a range of diseases that also require a protein restricted diet such as phenylketonuria (PKU), Maple Syrup Urine Disease (MSUD), and Urea Cycle Disorders (UCDs). Within the allied disorders, there are both national and regional support groups that help to connect families. If you or your family feel alone in adhering to diet, you may want to reach out to one of the family advocacy groups. HCU Network America's family conference is April 21 and 22 in Westford, MA. If Massachusetts is too far for you, some of allied disorders have conferences this summer in Colorado, Pennsylvania, Washington DC, and Georgia. Within New England, the Hole in the Wall Camp provides a place for kids to connect through summer camp. More and more, there are family support conferences, summer camps for children with rare genetic disorders, and specialty hospitals with community outreach programs.

This article is part of an ongoing series as part of the HCU and You: Tips and Advice. Share your questions or ideas for future articles at HCUNetworkAmerica@gmail.com

Sincerely,
Ben

Benjamin G Goodlett, PhD is a psychology postdoctoral fellow working with children and families affected by metabolic disorders.

Patient Clinic Survey



There are approximately 100 metabolic clinics throughout the US alone. One of the goals of HCU Network America is to connect with each clinic individually and establish a working relationship so we can help ALL patients and families affected by HCU.

Because there are so many clinics, we need your help! Please know your identity will remain anonymous! All entries received by March 31, 2018 will be entered in for $20 Amazon gift card raffle.

Please click the link and fill out the survey— https://hcunetworkamerica.org/patient-clinic-survey/

(Questions will include the following)
Name of Metabolic Clinic:
Clinic MAILING address:
Clinic Phone number:

Name of geneticist:
Geneticist email address:
Geneticist phone number:

Name of Dietitian:
Dietician email address
Dietician phone number:

Your name:
Your email address:

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 us helps us plan events, develop resources and educational tools, and to help better to insure 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/

Upcoming Events

Taking the Lead for HCU

Save the Date ~ April 21 & 22, 2018

The first HCU Network America Conference

Westford, Massachusetts

Haven't registered yet? [Register today]

Attention: Volunteers Needed

For HCU Network America's Conference: *Taking the Lead for HCU*
(An "educational meeting" for patients and families affected by homocystinuria, a rare genetic disease)

Westford Regency Inn and Conference Center | Westford , Massachusetts
April 21-22, 2018

We are now seeking a few adults to assist and help organize our children's program:

- Arts & Crafts
- Educational Lessons
- Stories
- Games
- Hands on activities
- And more!!!
- Compensation available depending upon experience

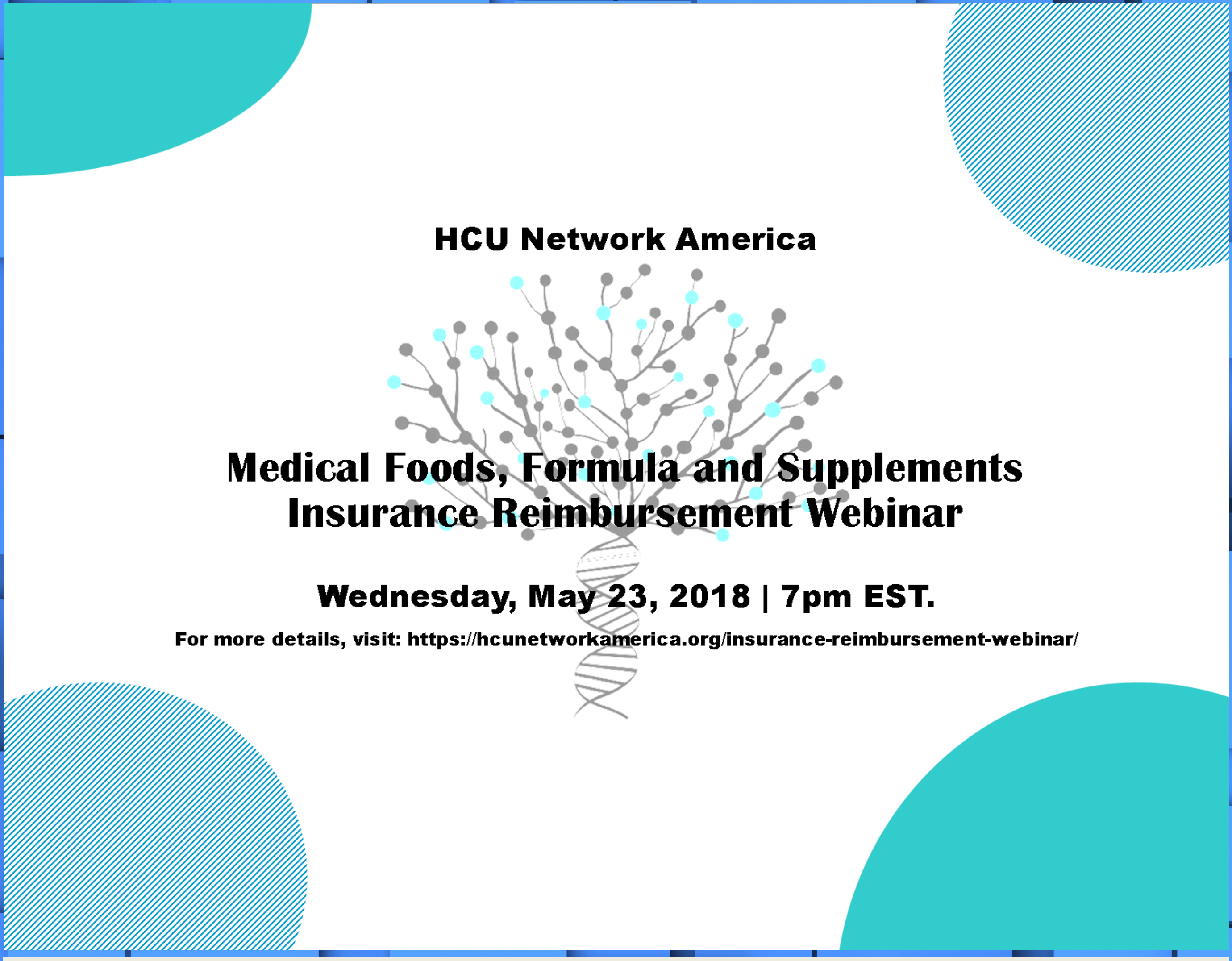
Children's program will take place during the main session of the conference.
(Saturday 8:00-5:00 and Sunday from 8:00-3:00)

Direct your contact with days and times of availability to:
Danae' Bartke, Executive Director at
hcunetworkamerica@gmail.com

Do you have a friend or family member who might be interested to assist in part of our children's program?

Please send them our way!

Upcoming Events, Cont.



HCU Network America

Medical Foods, Formula and Supplements Insurance Reimbursement Webinar

Wednesday, May 23, 2018 | 7pm EST.

For more details, visit: https://hcunetworkamerica.org/insurance-reimbursement-webinar/

During this webinar, Raenette will review information such as these coverage statistics:

- 50% of states have some type of insurance mandate.
    - Of those states, 84 % have some type of mandate on both food and formula coverage.
    - The other 16% percent of those states, do have mandates only on formula coverage.

*Some states may have programs that cover food or formula directly that are not included in these statistics.

Learn how you can get your food, formula and supplements covered by attending our FREE online webinar with Raenette Franco of Compassion Works Medical, LLC. Raenette is a Certified Biller Coder Specialist (CBCS), with over 17 years of experience helping patients get coverage on food, formula, and supplements they need!

To attend this webinar on May 23 at 7 PM EST, please register at:

https://hcunetworkamerica.org/insurance-reimbursement-webinar/

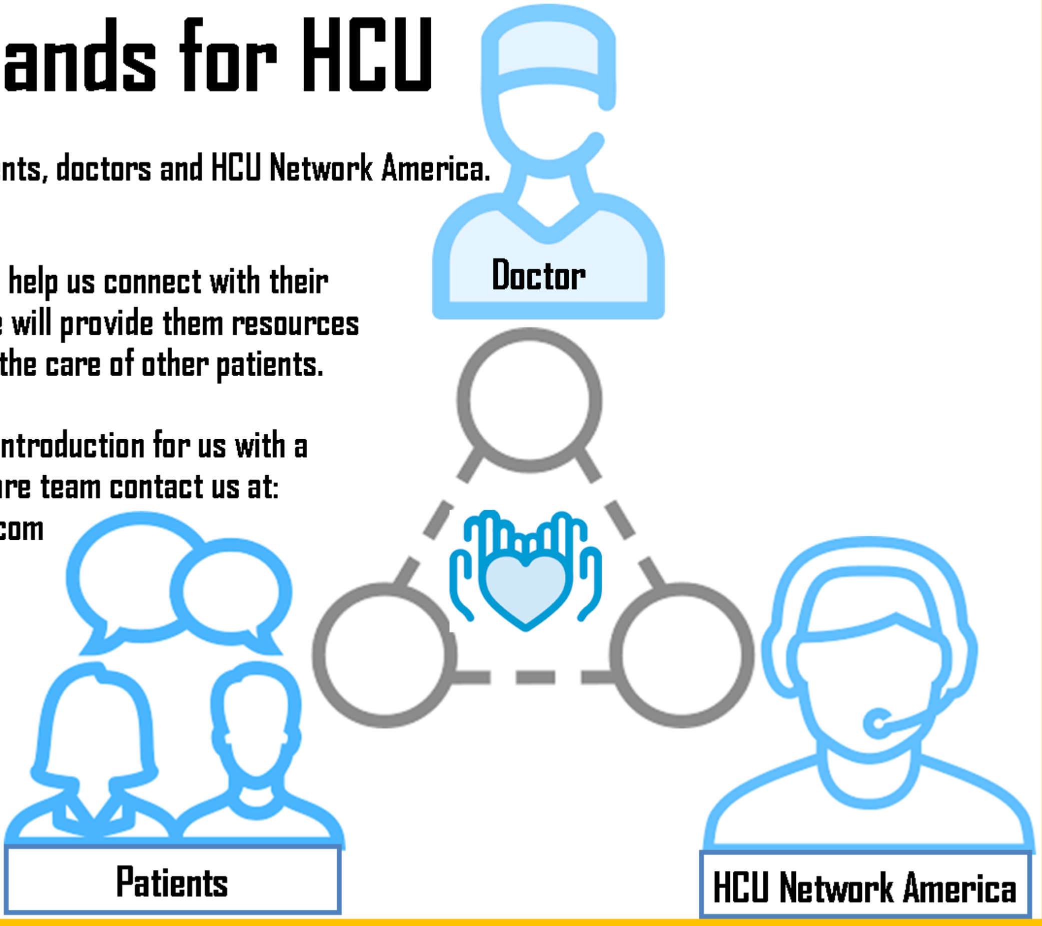
Ways to Get Involved

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.

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: Benjamin from Massachusetts
HCU and You: *Connecting the Dots:* Ben Goodlett