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:
Josie from Ohio

Josie is our third child and has been such a light in our lives since we found out we were pregnant with her. She was a beautiful baby and toddler, always making us laugh and such a sweet little girl. It was in her first few weeks of kindergarten that her teacher suggested something was going on, and perhaps she wasn't seeing the board as well as we thought. She already wore glasses so we had her eyes examined again. The optometrist referred us to Cincinnati Children's Hospital. After an eye exam there, we learned her lenses were slightly subluxated, indicating an underlying issue. About a month and a half later, Josie was diagnosed with homocystinuria, two week shy of her sixth birthday.

We were surprised at the diagnosis and the rarity of the disease. Her newborn screening card was pulled and her methionine levels had been within the acceptable range. We had never heard of a metabolic disorder. We are very fortunate to live 10 minutes from Cincinnati Children's which happens to be the number 2 children's hospital in the nation and houses a metabolic clinic within its Division of Human Genetics. Through our geneticist and dietician, we learned more about how to treat HCU. As her parents, we also took it upon ourselves to research it. We read every medical article on HCU, listened to every lecture and scoured the internet for any shred of information on this rare and often misdiagnosed condition.

No one we knew had ever heard of a low protein diet. In fact, this is the complete opposite of every societal trend right now. Who knew regular pasta and bread are loaded with protein? Fortunately, there are two other trends right now that greatly benefit the low protein community - - - gluten free and plant-based diets. Both are naturally lower in protein and methionine.

After the initial diagnosis, it took weeks of trial and error to learn that she was non B6 responsive. She has been on a low protein diet since January of 2018. She takes medication and supplements each day as well as vitamin B6. Every gram of protein she ingests is accounted for on a spreadsheet. We weigh and measure her food very carefully. Her diagnosis has meant making adjustments. For Josie, animal products are treated as a garnish. It takes creativity to adjust our family meals to make a version that is low protein. Our typical dinners are lasagna, tacos, pizza, grill out, pasta night, loaded baked potatoes etc. All of these, with the help of some medical foods and vegan options, can be made low protein and very tasty. She loves her vegan cheese, coconut milk and coconut ice cream.

Cincinnati is known for “Cincinnati Chili.” Our favorite is Skyline. A typical three-way (as we call it here) consists of spaghetti, secret recipe meat sauce (the Greek style chili) and loads and load of freshly shredded cheddar cheese. A regular size three-way has 45 grams of protein! Fortunately, our local Skyline lets us bring our low protein pasta, vegan cheese and even our own oyster crackers. We measure out the sauce for a delicious meal for her that is less than 10 grams. It’s Skyline time! (Shameless plug for our favorite chili - no we do not own one unfortunately). While we eat 90% of our meals at home, we do have pizza places that offer gluten free crust as well as Daiya vegan cheese which makes for a great low protein option.

Josie is becoming her own advocate. She excels at school, plays the piano, sings, does swim team and dance. We are confident she is the only child in her whole school who knows what methionine is and is ready to tell you. She is funny, sweet and confident. As we tell our children, many, many kids (and adults) deal with special issues on a day to day basis, whether it is food allergies, diabetes, autism, etc. HCU is one of these and as a family, we tackle it head on. Yes, it requires time and creativity but her blood results are worth it

The April 2018 HCU America conference was amazing. Making the connections with other families was invaluable. We are so excited for the next conference and will be attending as an entire family.

As we are coming up on one year since her diagnosis, our overwhelming feeling is one of gratitude to God for all the blessings since this diagnosis. We are thankful we got the diagnosis when and how we did. We are grateful for her excellent team at Children's. We are grateful for low protein food producers and her medication and supplement producer. We are grateful for Margie McGlynn, Danae' Bartke and the entire HCU Network America board. The timing of the inception of this organization and its conference relative to Josie's diagnosis can only be termed providential. And of course, we are grateful for Josie, who makes us smile and laugh every day and is always willing to try a new food.

Our advice to newly diagnosed families is this:

- Equip yourself with as much knowledge on this as possible.
- Reach out and use the support of the HCU America community. You are not alone!
- Give yourself some time to figure out how to navigate the diet, the medicine, etc. It takes lots of trial and error and patience.
- Help your child to become a self-advocate.

HCU Haberdashery

Official Merchandise of HCU Network America

Looking for the perfect Christmas Gift for your loved one with HCU, or one of those who looks after and advocates for them? Check out HCU Network America's official Merchandise Store, HCU Haberdashery. Proceeds from purchases benefit HCU Network America.

Get your HCU gear now, click here

HCU and You: *Recipes from the Kitchen*

Christmas Mint Cookies

Author: Amber Gibson

Yields about 36 Cookies
Protein: 0.27g protein per cookie
Calories: 105 calories per cookie

Ingredients:

290 gm (2¼ cups) wheat starch
1 teaspoon xanthan gum
50 gm (1/2 small box) jell-o instant pudding, vanilla flavored
1 teaspoon baking soda
½ teaspoon table salt
12 tablespoons (1 ½ sticks) unsalted butter, softened
½ cup granulated sugar
½ cup brown sugar
25 gm (1/2 large) mixed egg (egg white and yolk mixed together)
15 gm (1 tablespoon) water
1/2 teaspoon peppermint extract
5 drops green food coloring
45 gm (1/3 cup) semi sweet chocolate chips
50g Andes mint pieces

Directions

Heat oven to 375 degrees. Line two large baking sheets with parchment paper.

Whisk wheat starch, xanthan gum, pudding, baking soda, and salt in medium bowl; set aside.

In standing mixer fitted with paddle attachment or with hand mixer, beat butter, granulated sugar and brown sugar at medium speed until light and fluffy, about 3 minutes, scraping down sides of bowl with rubber spatula as needed. Add egg and water; beat at medium speed until combined, about 30 seconds. Add dry ingredients in two intervals and beat at low speed until just combined, about 30 seconds, scraping down bowl as needed. If the dough is a little dry, you can add water one tablespoon at a time. Add the peppermint extract and green food coloring and mix until food coloring is well blended. Add the Andes pieces and chocolate chips, stir until combined.

Form dough into tablespoon sized 1 -inch balls. Place dough balls on parchment lined baking sheet. If desired, freeze the dough balls on the baking sheet. Once frozen, place in a Ziploc bag for easy delicious homemade cookies in minutes.

Bake until cookies are golden brown around edges and just set and very lightly colored in center, about 12 minutes. If using frozen dough balls, cook for an extra three minutes. Cool cookies on baking sheet about 3 minutes; using wide metal spatula, transfer cookies to wire rack and cool to room temperature.

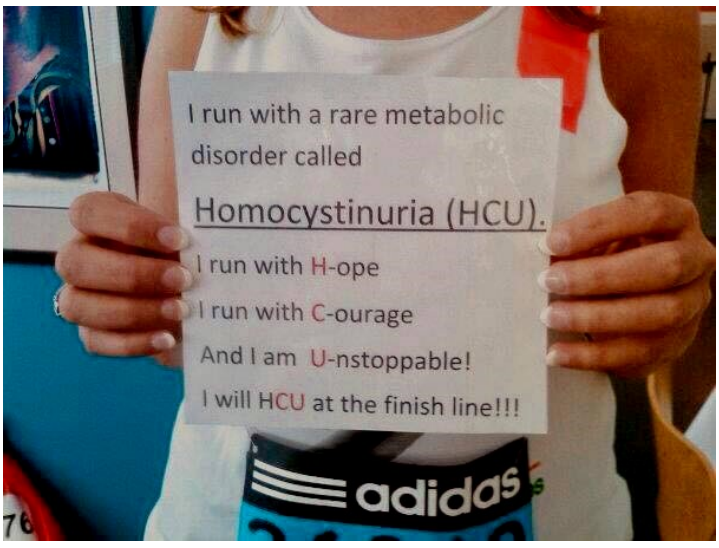
New News

The Rare Runner Updates

Stay Connected: the Rare Runner Blog

January 27, 2019 HCU Network America's Vice President, the Rare Runner, Kristin Rapp will run in the 2019 Fitbit Miami Marathon. This event will mark Kristin's 13th full marathon! As with previous marathons, Kristin will be bringing back her blog. Keep up to date with her thoughts and goals leading up to the 2019 Miami Fibit Marathon by following her blog.

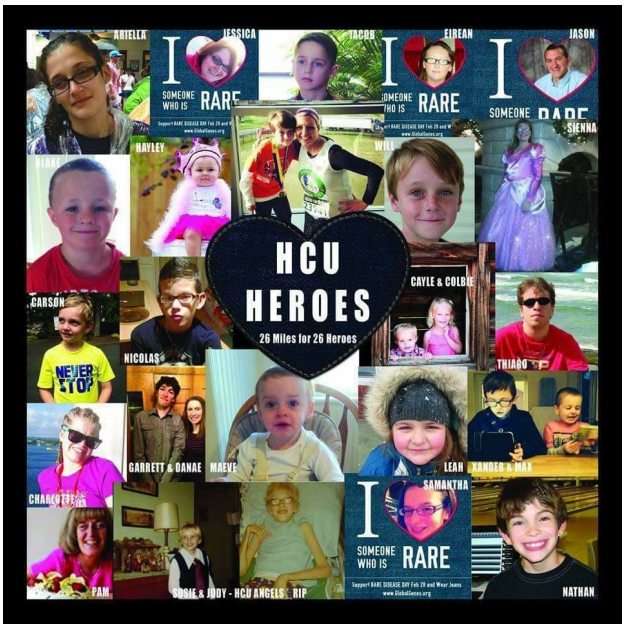
https://therarerunner.blogspot.com/



Get Involved:
Rare Runner Seeks 26 HCU Heroes!

Kristin is looking to dedicate each of the 26 miles to a HCU Hero. What's involved? Kristin would in turn ask that you share your HCU story, cheer her on (you can do that from the Internet too), and help advocate and help her raise funds.

Interested? Complete the form: https://hcunetworkamerica.org/hcu-heroes-participation-form/



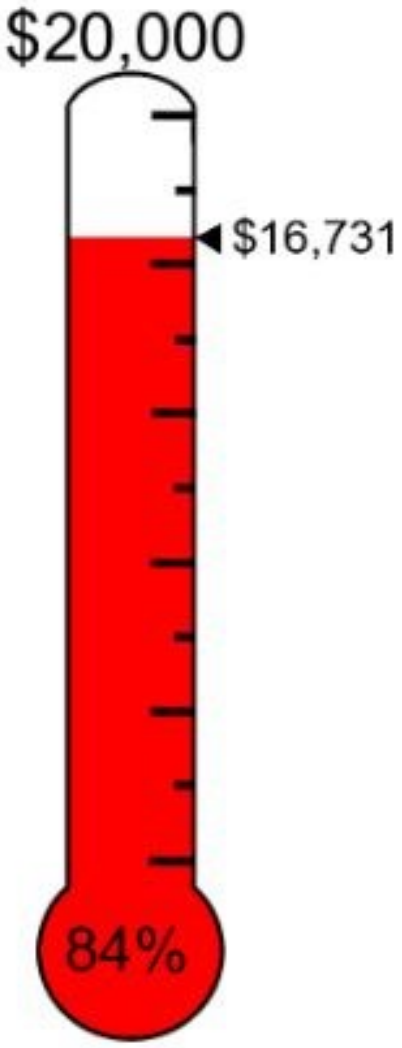
Donate!

Kristin has set an aggressive fundraising goal of $10,000 for HCU Network America. She is currently a little more than half way there. Help her reach her goal, donate today!

Click to Donate

2018 Fundraising Update

Thanks to an anonymous donor, any funds you help raise from October through December 31 will be matched up to $20,000!



Need information to complete your match?
Visit: https://hcunetworkamerica.org/company-

November Marks Beginning of
Open Enrollment for Health Insurance

Do you find your insurance coverage inadequate for low-protein foods, formula, betaine, or supplements? Don't fret—November marks the beginning of open enrollment for new health insurance policies.

Feeling overwhelmed? Not sure what policies cover your doctors and your medications? Don't worry, we can assist you with that!

HCU Network America has contracted Raenette Franco of Compassion Works Medical to assist you with your needs. Raenette can help you find a policy that works for you, or work with your current policy to help you get low-protein foods, medical formula, betaine and "supplements" covered.

There is no fee to work with Raenette, but we do urge you to contact her immediately if you do need a new policy. Open enrollment for 2019 ends December 15, 2018.

You may contact Raenette:
raenettef@compassionworksmrs.com
(973) 832-4736

2019 Rome Conference

See the link for more info: https://www.hcunetworkaustralia.org.au/patient-expert-meeting-2019/

HCU Network America Conference

HCU Network America will be holding its second patient/family-expert meeting October 19 and 20, 2019 at the Holiday Inn Indianapolis Airport. It will be a weekend full of friends new and old, networking, keynote speakers, breakout sessions, panels and a reception.

We listened to your feedback from last year and this one is very close to the Indianapolis airport. In addition each room has a refrigerator, microwave and individual cup coffee maker. The hotel has a 24 hour free shuttle that runs to and from the airport.

Discounted rooms will be available, so while we encourage you to save-the-date on your calendar, don't book your hotel rooms just yet. We will have a direct reservation link on our conference webpage.

Register your expression of interest: Click Here

OT-58, Enzyme Replacement Therapy Clinical Trial Recruitment

Orphan Technologies has initiated a first in human (Phase 1) clinical trial of OT-58, an enzyme replacement therapy that addresses the underlying enzyme deficit for patients living with classical homocystinuria. The goal of this trial is to evaluate the safety and efficacy of OT-58 in patients with classical homocystinuria and identify the appropriate dose. Patients between the ages of 12 and 65 years of age with classical homocystinuria may be eligible to join. For additional information on criteria for eligibility, please go to:

https://clinicaltrials.gov/ct2/show/NCT03406611?cond=Homocystinuria&rank=1

There are four sites in the US currently participating in the trial:

- Children's Hospital of Philadelphia – open to patient enrollment
- Boston Children's Hospital – open to patient enrollment
- Indiana University – open to patient enrollment
- Children's Hospital Colorado - will be open to patient enrollment in early Dec 2018

Payment for time and travel may be available to patients who participate in this trial.

To inquire about participation into the trial, please email: info@orphantechnologies.com

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/

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.

Newborn Screening Survey for Classical Homocystinuria

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

You may also fill and submit the form electronically by clicking here

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**

Formula Survey

Love your formula, hate your formula? Let us know exactly what helps you or stops you from taking it!

This short 5 minute survey will be used to work with formula manufacturers to develop and improve on current formulas' texture, smell and taste as well as develop formulas that better fits your life style.

Take the Survey Now

We'd like to thank the following content contributors:

Editor in Chief: Danae' Bartke
Heroes of HCU: Josie from Ohio
HCU and You: *Recipes from the Kitchen:* Amber Gibson

Click to donate directly