The HCU Herald

In This Issue

HCU Network America

Recipes from the Kitchen

Coconut Ginger Glazed Carrots

Ingredients
220 g Sliced Carrots
1 TBSP Butter, regular or unsalted
2 tsp Minced Ginger Root
2 tsp Sugar, Brown, packed
1 c Coconut Milk, canned
1/4 c Vegetable Broth
1 tsp Coconut Aminos
1 1/2 tsp Rice Vinegar

Directions

1. In a large skillet over medium heat, melt the butter. Add the sliced carrots and sauté for 5 minutes. Add the ginger and sauté another minute. Next, add the brown sugar and cook, stirring constantly, until the brown sugar has melted.

2. Add the coconut milk, vegetable broth, and coconut aminos. Continue to cook over medium heat until the carrots are fork tender. Some of the liquid will evaporate, leaving a nice, sweet glaze over the carrots. Add the rice vinegar and stir for about 1 minute. Serve hot.

Notes: If you are not a fan of ginger, you can reduce it to one teaspoon, or omit.
Servings: Yields 3
Serving Size: 95 g
Protein per serving: 1.8 g
Calories per serving: 191

Fruit Dip

Ingredients
4 TBSP Pea-Not Butter, softened
4 TBSP Cambrooke Cream Cheese
3 TBSP Sugar, Powdered (Confectioners)
1/2 tsp Vanilla Extract
1/8 tsp Salt, Table

Directions
Combine all ingredients in a small bowl and mix until combined. Refrigerate until ready to use.

Notes
For a nice fall treat add a little cinnamon and a dash of nutmeg.

Servings: Yields 5.4
Serving Size: 2 Tbsp
Protein per serving: 0.4 g
Calories per serving: 72

Savory Cheeseball

Ingredients

4 fl.oz. Cambrooke Cream cheese Garlic & Herb, softened
2 tsp Chopped sun-dried Tomatoes, packed in oil 6mg
14 g Mozzarella Shreds
1 clove(s) Garlic, fresh cloves, chopped
10 g Pretzel Sticks, broken in half

Directions

1. In a small bowl add the garlic herb cream cheese, chopped sun-dried tomatoes, mozzarella shreds, and garlic. Use a spatula to blend all ingredients together until well combined and smooth. Add salt and pepper if desired, mix again.

2. Pull out a piece of plastic wrap big enough to wrap the cheese filling tightly. Add the cheese filling to the plastic wrap and wrap tightly, forming a ball shape. Refrigerate for at least 4 hours to help the cheeseball hold its shape. I refrigerated mine for 2 days.

3. When ready to serve the cheeseball it is time to make the coating. Place the broken pretzels sticks in a small food processor and pulse until you have small crumbs. Try to not make it into a powder. The larger pieces will give a nice crunch. Take the cheeseball out of the fridge and gently coat with the pretzel crumbs. Serve with your choice of crackers or pretzels.

Servings: Yields 7.5

Serving Size: 2 tsp

Protein per serving: 0.4 g

Calories per serving: 37

News and Events

September is Newborn Screening Awareness Month!

Ways to get involved:
Share how newborn screening (or lack of) has impacted your life!
How do I share my story?

- Include the basics.
- Be authentic.
- Use images or video.
- Choose one story to tell.
- Try and include a call to action.

Where do I share?

- With HCU Network America
- Facebook and Facebook Live
- Baby First Test (https://www.babysfirsttest.org/newborn-screening/family-experiences)
- Twitter
- Instagram Live

Raise Awareness on Facebook –

- Post, share, and comment using the hashtag #2019NBS
- Add the #2019NBS frame to your profile picture

1. Update your profile picture
2. Click add frame
3. Search for #2019NBS
4. Reposition your profile picture accordingly
5. Determine how long the frame will be displayed

Raise Awareness on Twitter
See example Tweets

- Attend the twitter chat!

October is HCU Awareness Month!

This year's theme is Hope Connects Us. Why did we pick this theme? Some might say it's because Hope Connects Us starts with HCU, but that's not the official reason. Hope Connects Us was chosen because as NORD (National Organization of Rare Diseases) puts it, "alone we are rare, together we are strong!". Having a community to draw upon gives us strength, courage, and a voice that is much more amplified. So help spread hope and get connected with the Homocystinuria community this month.

October is packed with exciting things to come – here is a sneak peek. Mark your calendars and make sure you are ready!

Click Campaign
Get your mouse ready and be prepared to click! For one week only (October 1-7th) Recordati Rare Diseases will donate up to $5 (up to $5,000) to HCU Network America for every click. Be on the lookout for the link.

HCU Awareness T-Shirts are back!
Get your official 2019 HCU Awareness T-Shirts. All proceeds benefit HCU Network America! Order your shirt now so you have it in time for HCU Awareness Month! Click a shirtto purchase !

Buy me now:
bonfire.com/hope-connects-us-letter-tiles
Pick me, Pick me!
bonfire.com/hope-connects-us-hands-together

HCU Heroes

Gabbi

From Massachusetts

Hi, my name is Gabbi, and I was the first baby diagnosed with HCU by amniocentesis. Benjamin, my older brother, who also has homocystinuria was diagnosed through newborn screening. My sister, Chloe, on the other hand, is fortunate to not have the genetic condition. Knowing that I had HCU before I was born was helpful because I was able to drink the specialized formula as soon as I was born, for that I am truly lucky. Although, just because I've been on formula and a restricted diet my whole life doesn't mean I haven't had my fair share of complications. At age 8, I was diagnosed with scoliosis. I was put into a tight, restrictive brace just hoping it would work enough to avoid surgery.

For six years I was seeing an orthopedic doctor at Tufts Floating Hospital. It wasn't easy wearing the brace, and I ended up with a total of 5 different ones in that 6-year period. In June of 2016 my doctor gave me the news that I needed to have spinal fusion surgery to correct 3 intense curves. For a person with just scoliosis, the surgery would be tough, but not life threatening. For an HCU patient, surgery can be dangerous. Instead of giving up, my parents and I sprung to action. We transferred to Boston Children's Hospital to be with my metabolic team and saw an orthopedic surgeon in the next following weeks. I ended up having surgery just a couple months later on August 4, 2016. Because of the potential complications of having surgery, I went in the night before for extra fluids to help diminish the risk that HCU poses. I never would have expected that week to go how it did. It consisted of 7 days in the ICU and definitely didn't go smoothly. The months following were filled with pain and healing. Six months passed and the pain just never went away. One year passed and the pain was worse. I would always go back to my surgeon and tell him that I was still in pain and I'd always get the same response about my X-rays being normal. I knew something was very wrong, but I felt the doctors weren't truly listening.

I was becoming discouraged, and I just never felt myself anymore. This is when I decided to find an outlet. That happened to be photography. I never realized how helpful photo imagery would be through this trying time in my life. I entered The Rare Artist Contest in 2017. My entry was entitled, "The light in the dark." It reflected how one might be challenged to see the positivity in life and the need for one to grasp for and hold onto their light. As much as I had withstood intense pain throughout the past couple years, I never permitted my physical issues to impede my spirit. I remained hopeful despite my circumstances. My goal in entering the contest was to inspire others with rare disorders. Little did I know that my entry would win. I was invited to Washington D.C. to speak on Capitol Hill in front of hundreds of people about living with a rare disorder and my love for photography.

HCU Hero: Gabbi

Winning the Rare Artist Contest was an incredible honor. Sharing my story in front of members of Congress and influential individuals, along with others with rare disorders, was empowering. It provided me with a platform for exposing rare diseases and the complications endured to those who may have had little knowledge of Homocystinuria.

After D.C. I decided to advocate for myself and find out the cause of my pain. More recently I have switched surgeons, got a CAT scan, an MRI, and many more x-rays. Truly what is going on could be a variety of complications resulting from my surgery and not just one clear answer on how to fix them. We have gone ahead and scheduled a surgery date in December of 2019 to completely redo my fusion. I hope in the time between now and then the doctors figure out what's going on and that surgery isn't the answer, but there's no promises. Although I know I can get through another surgery, the potential for infection worries me the most.

I have gone through a lot in life and it has been hard, however it has never stopped me. I'm going into my senior year and applying to fantastic colleges. I have taken the SATs 2 times now and have gotten great scores. I am taking 2 AP classes and 3 honors classes this upcoming year. I'm the President of Peer Leaders, I'm in the English Honors Society, and I work as a teacher aid. HCU has never stopped me academically. In fact, I have excelled in all my classes. My pain, while disruptive, has never stopped me either. I see it more as a piece of me. Being in hospitals a lot I have learned I want to work with children. Continuing on with that I want to be an elementary school teacher and inspire kids to realize that they can achieve anything. I want young children to know that there is nothing they cannot do and to always think outside the box, dream big, and to treat others with kindness because you never know what they're are going through.

HCU Hero: Gabbi

When I was asked to write this, I knew I didn't want to just tell my story. I wanted to show parents with young children that HCU will never stop them. Yes, it is a challenge, but it will only limit them if they let it.

Lastly, to my brother Ben, you truly are my role model. You are successful and a pioneer for the HCU community. I love you so much and remember, take your medicine :). To my sister, Chloe, you are my best friend. You've taught me to think positively and to never let anything stop me. To my parents, thank you not only for being the supportive loving parents you are, but for dealing with my teenage attitude. Finally, to whoever is reading this, whether you are a parent, sibling, friend or family member of a child with HCU, being rare is beautiful, never forget that.

Are you the next HCU Hero ?

A Hero is an ordinary individual who finds the strength to persevere and endure in spite of overwhelming obstacles.
– Christopher Reeve

HCU Awareness Calendar

Want to do more to raise awareness for HCU? Check out our fun online activities you can take part in to help those understand more about HCU.

HCU Fact Calendar
This year we are spicing things up and working to raise more awareness than ever! This year we won't just be sharing facts about Classical Homocystinuria, but also the other homocystinurias which include the Cobalamin Defects and Severe MTHFR.

Week 1 – General HCU Awareness
Week 2 – Classical Homocystinuria Facts
Week 3 – Cobalamin Defects
Week 4 – Severe MTHFR

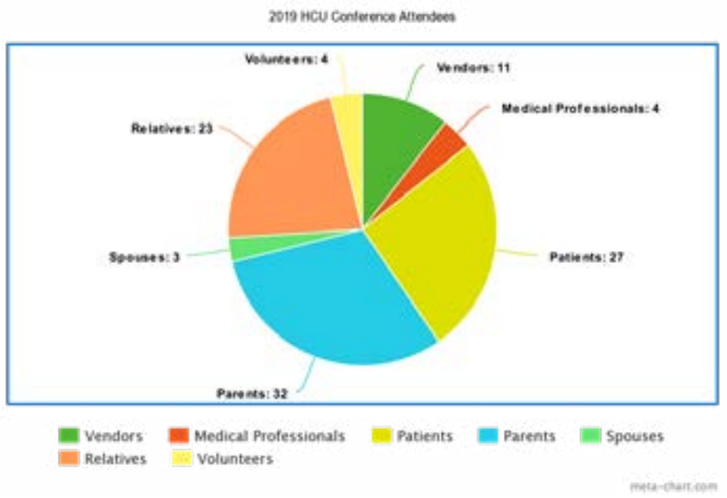
October 2019

Sun	Mon	Tue	Wed	Thu	Fri	Sat
		1 Change your Social Media picture to the HCU Awareness Ribbon	2 Start your HCU Facebook Fundraiser and Invite your friends	3 Share the Classical HCU infographic OR Cbl pathway and share there are 10 types of Cobalamin defects!	4 Share a HCU patient story! Videos: http://bit.ly/PatientStoriesVideos Text: http://bit.ly/HCUHeroes	5 Share your diagnosis story!
6 Challenge your friends to eat 10 g of protein and a normal protein shake 3 times a day. #ToastToHCU	7 Share a pic of an item that has the same amount of protein you can have	8 Share your daily diet record - completed	9 Share a low protein meme	10 Share your favorite low protein recipe! Bonus if you cook it and share a pic.	11 Dining out, low protein style	12 Share a pic of what your grocery store "haul" looks like
13 Real Coast of HCU: Grocery Cost Comparison #MedicalNutritionEquityAct OR Some patients with HCU require injectable B12. B12 on average is $300-400 a month and most insurance companies don't cover it!	14 Share a picture or video capturing all the medication, or of you taking your medication for the day (this includes formula for those who need it).	15 Share a pic of your first pair of glasses - or a device that helps you navigate or communicate due to lack of vision	16 Share something you wish people would understand about HCU	17 #HaikuforHCU Write and share a Haiku describing life with HCU	18 Wear jeans for your rare genes	19 Wear your HCU Shirt and share a pic online
20 #GoBlueForHCU	21 #HCUAwareness post in a public place	22 Share with stranger what HCU is and why it's important to you	23 #Create4Cure create a work of art that brings awareness for HCU	24 #High5forHCU List 5 ways HCU makes you a stronger, better person	25 All states test for Classical HCU, but many patients are still missed	26 Share a picture of you and a HCU buddy! Or tag a friend who is a great support!
27 #FacesOfHCU Share a picture of you saying - I am one of the estimated 1 in 200,000 people with HCU	28 #Hope4HCU Share 4 things that give you hope and encouragement	29 Share the HCU timeline - if you know other facts, let us know!	30 Cutting Edge of HCU: Share about a therapy that is in the works!	31 Your message to the world about HCU!		

print-a-calendar.com

Get your engines ready

Join us in Indy for HCU Network America's 2nd Homocystinuria conference, Accelerating Towards a Cure. Join the 100+ attendees from 22 states. Registration runs till October 4, 2019. Discounted room rates end September 18, 2019.



Want to get more involved, but don't qualify for the Enzyme Replacement Therapy Trial – Drive research by joining the Natural History Study!

Current sites include: Atlanta, Boston, Denver, Indianapolis and Philadelphia. 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.

Click here to learn more

VMP, LLC
expanding genetic horizons...

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HELP US TEACH PHYSICIANS ABOUT HOMOCYSTINURIA

FACT! Teaching about metabolic diseases in medical school and residency programs is poor.
FACT! Most patients live and die without a diagnosis being made, especially when the disease presents in adulthood.
FACT! Patients cannot access effective therapies unless a proper diagnosis is made.
FACT! The sooner a diagnosis is made and treatment begun, the better the outcome.

WE NEED YOUR HELP!

We at VMP Genetics believe in the power of "patient-teaching" and are bringing patients and families into lectures and presentations - at conferences and in the classroom around the country. While doctors teach facts, patients tell stories. Story-telling is a more compelling teaching method with better recall over time than didactic lecturing. We also believe that doctors are more likely to make a diagnosis if they have already seen a patient and heard her/his story. Story-telling can be live or taped...

WE ARE LOOKING FOR...

- ***Patients and/or family members who are interested in telling their stories in local medical classroom settings...*** We are developing a Patient Teacher Registry. If a medical school faculty member is looking to introduce the patient story in a teaching session, the Registry can tell him/her if there are patient-speakers in the area and what diagnoses they have.

- ***Patients and/or family members who are interested in having their stories videotaped...*** As we secure funding, we are interested in recording stories that reflect the broader patient experience. The more variety in the stories, the richer the learning potential.

- ***Videos of patients and families telling their stories...*** A 5-or 10-minute clip can be downloaded into a lecture about that disease or relevant biochemistry to enhance the learning potential of the session.

Please help us in our efforts to raise awareness about Homocystinuria through this innovative educational outreach to the medical community. To Volunteer or participate, or for more information about this project... please contact Jacob Athoe at PatientTeacherRegistry@vmpgenetics.com

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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 defect 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 - open to patient enrollment
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/
https://hcunetworkamerica.org/contact-register/

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HCU and You: Recipes from the *Kitchen:* Amber Gibson

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 viae-mail at:
FICICIOGLU@email.chop.edu

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

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