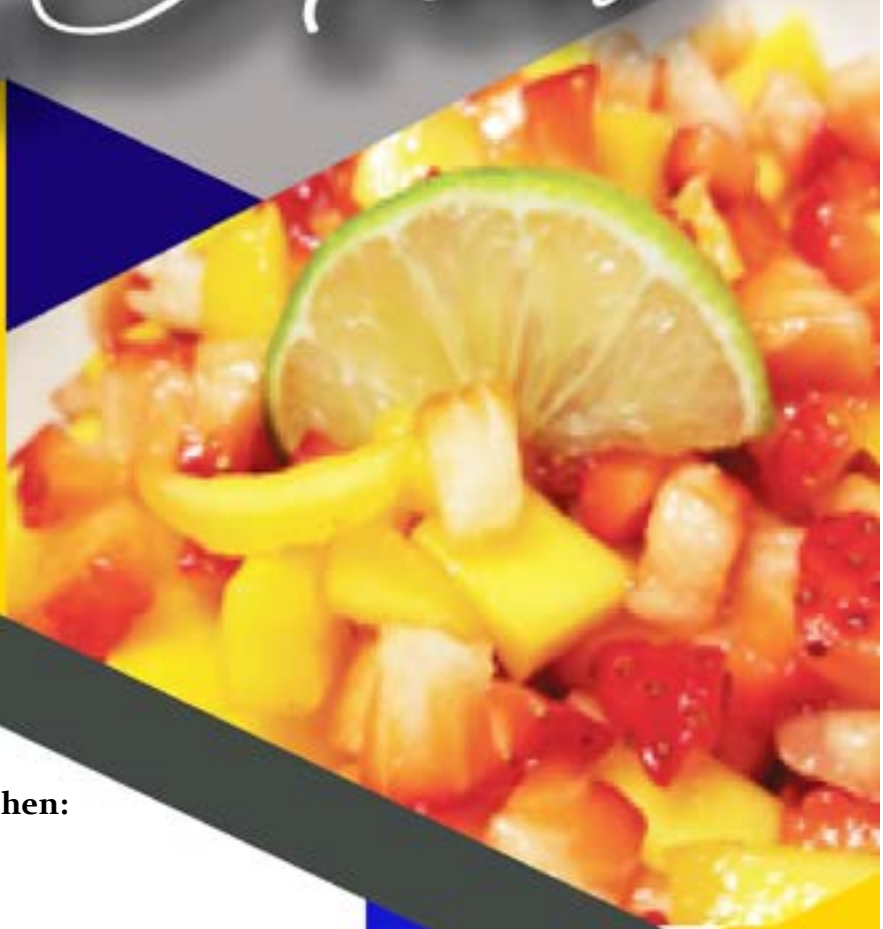
The HCU Herald



In This Issue

HCU Heroes: Jessica from Michigan

HCU and You Recipes from the Kitchen:
Strawberry Mango Salsa
Buffalo Jackfruit Dip
Pesto Pasta
Low Pro Cannoli

Register Now:

Accelerating Towards a Cure 2019 HCU Conference

Be Your Own Advocate Webinar

Back to School with HCU Webinar

New News:
Solace Nutrition Ad

Fundraising Tip

Get Involved:
Newborn Screening Survey
Natural History Study

Connecting for a cure. There have been great things happening for the HCU Community and HCUNA. We strive to keep you informed and connected.

HCU&You:Recipes from the Kitchen

Strawberry Mango Salsa

Ingredients:

145 g strawberries, diced

100 g mango, diced

1 TBSP. Lime Juice

½ tsp. Juice from jar of pickled jalapenos

¼ tsp. salt, table

½ tsp. Cilantro, raw, chopped

Directions:

Combine all ingredients in a glass bowl and lightly toss to combine.

Cover with a lid and refrigerate until ready to use.

Servings: 5.3

Serving Size: ¼ cup

Protein Per Serving: 0.3 g protein

Calories Per Serving: 22 calories

Cannoli Filling

Ingredients:

130 g Cauliflower, raw florets, steamed and cooled

60 g Cambrooke Cream Cheese, room temperature

1/2 tsp Vanilla Extract

1/4 c Sugar, Powdered (Confectioners)

1 tsp Orange Peel (zest), fresh

2 tsp Orange Juice, fresh

1/2 c CocoWhip Coconut milk Thawed Frozen Dessert Topping, lightly packed

30 g Semi-Sweet Chocolate Mini Chips

Serve with low protein graham crackers, or in a low protein cannoli shell.

Servings: 5.7

Serving Size: ¼ cup

Protein per serving: 0.7 g per serving

Directions:

In a food processor with a fitted blade, add the cooked cauliflower with the cream cheese. Pulse until texture resembles ricotta cheese, about six times. Scrape in between pulses.

Add the vanilla, powdered sugar, orange zest, and orange juice. Pulse to combine. Pour into a medium bowl. Add the coco-whip and gently fold just until combined. Add mini chocolate chips and fold into the filling. Refrigerate for 4 hours or overnight. Filling is ready to use.

Nut Free Pesto Sauce w/pasta

Ingredients:

70 g Spinach, fresh, torn

20 g Fresh Basil Leaves

4 TBSP. Olive Oil

30 g Parmesan Style Cheese, Shredded

Directions:

Place the spinach, basil and two tablespoons of the olive oil in a food processor. Pulse until the spinach and basil is finely chopped. If it seems a little dry, add the last two tablespoons of olive oil and pulse until combined. Next add the Parmesan cheese and pulse just until combined.

Servings: 15.5
Serving Size: 2 TBSP
Protein Per Serving: 0.2 g protein
Calories Per Serving: 39 calories
Methionine per recipe: 46 mg

To store, place pesto in a glass jar fitted with a lid. Refrigerate up to a week or freeze up to 3 months. You can also freeze in ice cube trays to make portions easier to use.

Nut Free Pesto Pasta:

Ingredients:
75 g Aproten Linguine
1 TBSP Cambrooke Alfredo Sauce Mix
1 TBSP Butter
1/3 c. non-dairy creamer
1.5 tsp Nut – free pesto (above)

Directions:

Cook pasta according to directions and set aside
In a small skillet add the creamer and 1 TBSP butter. Place skillet over medium heat and heat until butter melts.
Add the alfredo mix and lightly whisk until sauce thickens.
Add nut-free pesto and whisk to combine.
If sauce is to thick, add a little more creamer until desired thickness is reached. (Adjust protein content)
Toss pasta in the sauce and serve immediately.

Servings: 2
Serving Size: 155 g
Protein Per Serving: 1.3 g
Calories Per Serving: 130 per serving

Buffalo Jackfruit Dip

200 g Young Jackfruit in Brine, drained, seeds removed and roughly chopped

¼ c. Franks Buffalo Sauce

8 fl. oz. Daiya Cream Cheeze, Plain, room temperature

2 TBSP. Sour Cream

2 TBSP. Rice Dream, Original

¼ tsp. Garlic Powder

¼ c. Daiya Cheddar Style Cutting Board Shreds

¼ c. Daiya Cutting Board Mozzarella Style Shreds

In a glass or ceramic bowl, add the chopped jackfruit and buffalo sauce. Gently toss to combine. Cover and place in the fridge to sit for 30 minutes.

In a medium sauce pan, add the cream cheeze and cook over medium low heat. Stir frequently to prevent burning. Once melted, add the sour cream, rice milk, and garlic powder. Continue to cook and stir over medium low heat until the cream mixture begins to bubble.

Remove the buffalo jackfruit from the fridge and toss into the creamy mixture and lightly mix. Turn heat up to medium and add the cheeses. Stir until melted and thickened. Serve hot with tortilla chips and crackers.

Chips and crackers are not included in the overall protein information. If you want this dip more mild, reduce the buffalo sauce to 2 tablespoons

Servings: 6.63

Serving Size: ¼ cup

Protein Per Serving: 1.1 g protein

Calories Per Serving: 139 calories

HCU Heroes

Jessica

From Michigan

The Early Years:

I was diagnosed with homocystinuria at the age of 4 and a half. I don't remember much before I was diagnosed other than I loved chicken. As a child, I was devastated that my life was going to change forever. I soon realized that my life would consist of many doctors' appointments, and I would be missing a lot of school. I wanted to be normal and did not know how that would be possible.

Once diagnosed, I was sent to the pediatric geneticist at University of Michigan. My diet changed almost immediately; I started taking a formula which had all the amino acids in protein except methionine. I also had to eat a low protein diet. As a child, I didn't understand why I was different. Friends would often ask, "Do you want a candy bar?" Initially, I would quickly respond to this question saying, "I'm allergic." I now explain that my body does not process protein effectively.

We spent the first six years at the University of Michigan, until my doctor announced that he was going back to Baylor, Texas. He recommended a specialist out of Rainbow Babies in Cleveland, Ohio. For the next few years I continued my care there.

Camp

When I was about 12 years old, I finally met someone who lived in my state and also had Homocystinuria. It was very hard to keep in touch because of the distance between us, but I finally realized I was not alone. Around that year, I started going to a PKU Camp in Ohio. I learned there are other people who have metabolic disorders, and they also have to follow a low-protein diet. Throughout the years, between the summers, I often felt like no one understood me. I was constantly explaining myself and answering hundreds of questions. This was overwhelming as a teenager who didn't have a lot of communication with other HCU patients.

The College Years & Beyond

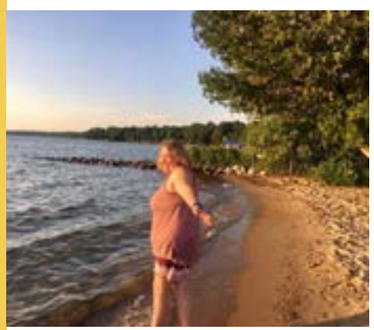
Right before my 21st birthday I switched doctors to the Detroit Medical Center where I would be for the next 5 years. At first was happy, but I grew bitter against the physician as I was lied to on multiple accounts. At the same time, Michigan's governor passed a law stating that if you have HCU you would qualify for Medicaid and the formula would be covered, but not the low protein food.

I was able to go back to Mott Children's Hospital. I am now under the care of my original team who is honest and is teaching me more as an adult living with HCU. One thing about living with HCU is remembering support is key. I am very lucky to have a great supportive family who has encouraged me to live my life to the best of my ability. My best piece of advice is to stick close to the ones who support you. My parents are my rock. They've been the most supportive people in my whole life.

Formula & Treatment

Michigan didn't have many choices for HCU formulas. The first formula I was prescribed was Hominex 2. I was on that formula until I was a junior in high school. During my junior year, my nutritionist at the time, encouraged me to switch to the Vitaflo HCU coolers. I was on the coolers for about 8 years, until my body developed some allergic reactions to them. I am now on Mead Johnson HCY2. Unfortunately, it doesn't have the convenience of being pre-mixed. I think it smells like cake batter, and boy do I wish it tasted like it. Now I'm currently trying HCU Easy Tabs.

Treatments are always changing, and I am thankful for that. My prior metabolic team had asked me to consider a liver transplant. I wasn't comfortable with this because there wasn't a lot of research done on it. Deciding to wait it out was the best decision I could have made. In January of 2019, I started my enrollment in the clinical trial of OT-58. This drug looks promising for the HCU community. I'm looking forward to seeing how this drug can shape HCU history.

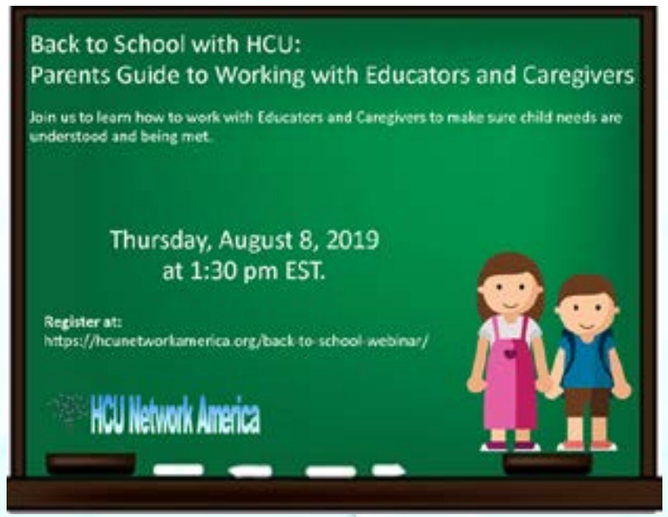


Are you the next HCU Hero ?

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

Register Now !

Register Now!



Get Involved

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 patient's 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

Contact VMP

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

VMP Genetics
Contact Register

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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:

We'd like to thank the following content contributors:
Editor in Chief: Danae' Bartke
Heroes of HCU: Jessica from Michigan
HCU and You: Recipes from the *Kitchen:* Amber Gibson

Natural History Study

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, 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. Learn More !

Accelerating Towards a Cure
2nd Homocystinuria Conference
October 19-20 2019

Our conference map is filling up! Conference attendees represent 16 states and spreading! Let's light up our map blue for HCU – register now at https://hcunetworkamerica.org/2019-conference

Charity Rummage Sale

Plan:

1) Recruit volunteers—even if you think you can do it alone, start recruiting help; you will need it!
2) Pick a weekend
   - Don't pick a holiday weekend. Holidays will tend to be slower and limits your volunteer availability.
   - Check your city calendar for big events, these can help and hurt you, depending on the location of your sale.
   - Check availability if holding the sale at a church building, community center or parking lot.
3) Choose a location:
   - Try to aim for an area with a lot of visibility, easy traffic and parking
     - Places to consider:
       - A church
       - Rec center, or community center
       - School gymnasium
     - Does your place allow for a back up plan?
       - Can you move inside if it rains?
       - Can you set up tents if it rains?

Collect:

1) Collect donations—
   - Start a minimum of 6 weeks in advance
     - Ask people to gather unwanted items via word of mouth, flyers, emails and/or social media
     - Designate a drop off locations, dates and times.
     - Offer to pick up items for elderly or disabled peoples
   - Have people collect plastic grocery bags—loads of them!

Advertise:

1) Let people know why you are raising money
2) Spread the word on social media
3) Set up a Facebook Event Page—add a map!
4) Advertise on Craigslist
5) Put up flyers

Sort and Sell:

- Sort and price items—start once items start to come in.
  - Price things low, so they sell!
  - Use color coded dots , ex. Lime green 25 cents, blue 50 cents, red $1 etc.
  - Make signs detailing the color coded system for dots, or stickers
  - Clothes
    - Sell clothes by the $5 per bag
    - Hang nicer clothes (coats, formals, business wear) price individually
- Put unpriced and unorganized items in a place separate from the sorted and priced
- Set up signs advertising and directing people to the sale
- Arrive early on sale day to set up. If possible, set up day or two in advance
- Have a couple people great shoppers and explain "how it works" and the cause (be brief).
- A couple hours before end of sale ends, sell things in bulk prices—$5 per bag or 50% off.
- Allow people to haggle
- Pack and Clean up
- Decide what you will do with items that don't sell: donate, send home, use for another sale.