The HCU Herald

HCU Hero
Rileigh from Ohio

Fundraising Inspiration:
March Madness: March 17, 2020
Pub Crawl

Recipes from the Kitchen:
Cucumber Salad
Shepherd's Pie
Key Lime Pie

Texas Event
NBS Survey
Contact Register

HCU Network America

Recipes from the Kitchen w/Amber Gibson

Cucumber Salad

Ingredients
620 g Peeled and thinly sliced cucumber, about 2 large cucumbers
2 tsp Salt, Table
12 g Diced white or yellow onion
2 fl.oz. Cambrooke Cream Cheese, room temperature
1/4 c Sour Cream
1 tsp Lemon zest, fresh, about one lemon's worth
4 tsp Lemon Juice, about one lemon's worth of juice
5 sprig(s) Dill Weed, fresh, finely chopped

Directions

1. Place the thinly sliced cucumbers into the colander. Add the 2 teaspoons of salt to the cucumbers and gently toss. Allow to sit for 30 minutes.

2. In a medium bowl add the onion, cream cheese, sour cream, lemon zest, lemon juice, dill, and sugar. Gently whisk to combine and set aside.

3. After sitting for 30 minutes, pat the cucumbers dry with a paper towel. You do not want too much moisture in the final product. Add the cucumbers to the sour cream mixture and gently toss to coat. Place into a container with a lid and seal. Refrigerate until cold, or up to overnight to allow flavors to meld. Serve cold.

Notes:
Yields: 8 servings
Serving Size: 82.5 g
0.8g protein per serving
36 calories per serving
Serving Size: ¼ cup

*The protein and PHE values are estimates and based on using regular mashed potatoes and tomato paste.

Yields 8 servings
Serving size: 125g
1.6g protein per serving

Vegetable Shepherd's Pie

Ingredients

48g Diced Onions (0.5)
2 Cloves Garlic, Minced (0.4g)
75g Diced Carrots (I actually used carrot spirals) (1.1g)
100g Diced Zucchini (1.2g)
100g Diced Yellow Squash (1.2g)
75g Diced Parsnips (0.9g)
50g Chopped Mushrooms (1.6g)
70g Diced Tomatoes (0.6g)
1 TBSP Tomato Paste (0.7g)
1 cup Veggie Broth (0.4g)
¼ cup Sherry (optional)
1 packet G Washington's Rich Brown Seasoning and Broth mix (0g)
2 Tbsp Balsamic Vinegar (0g)
One Sprig Thyme
One Bay Leaf
One Sprig Rosemary
2 tsp corn starch mixed with 1Tbsp water

Directions

1 cup (328g) Mashed Sweet Potatoes (4.4g) or 1 cup (248g) mashed potatoes (4.8g)

1. In a large skillet over medium heat, add 2 tablespoons of olive oil. Once oil is heated and glistening add the onions and sauté until translucent. Next add the garlic and sauté for one minute.

2. Next, add the carrots to the skillet. You can add a little more olive oil and necessary. Cook, stirring constantly, until the carrots begin to soften- about 5 minutes. Add the zucchini, yellow squash, and parsnips. Continue to cook for another 4 to 5 minutes. Toss the mushrooms and tomatoes into the skillet and sauté for one minute.

3. Add the tomato soup, vegetable broth, sherry, seasoning mix, balsamic vinegar, thyme, bay leaf, and rosemary. Bring to a boil, then reduce to a simmer. Allow the filling to simmer until vegetables are tender. Add the cornstarch combined with water to the filling and simmer for one minute to allow the gravy to thicken.

4. While the filling is simmering on the stove, preheat the oven to 375 degrees Fahrenheit. Spray a medium casserole dish with cooking spray.

5. Pour filling into the casserole dish. Top with prepared mashed potatoes of choice. You can use a spoon to make a decorative pattern with the potatoes.

6. Bake in the preheated oven until everything is heated through and the potatoes have browned a little, about 15 to 20 minutes. Remove from oven and allow to sit about two minutes. Serve and enjoy!

Key Lime Pie

Ingredients

Crust:

180 g Kinnikinnick Vanilla Wafers, 1 full box 370mg

4 TBSP Butter, regular or unsalted, melted 24mg

Key Lime Filling

13 2/3 fl.oz. Coconut Milk, canned, 1 full can 176mg

50 g Lemon Pudding, dry mix only

1/4 c Sugar, White Granulated

2 tsp Agar Powdered 15mg

1/4 c Lime Juice 6mg

1. Preheat oven to 350 degrees. Place vanilla wafer cookies in a food processor with a blade attachment and pulse until you have crumbs, as if making a graham cracker crust. Pour crumbs into a small bowl and add the melted butter. Gently toss to combine. Pour mixture into a 9 in pie tin. Using the flat bottom of a glass, press the crumbs into the pie tin, creating a nice edge evenly around. Bake for 14 minutes. Remove from oven and allow to cool completely. Turn oven off.
2. In a small sauce pan, add the canned coconut milk. Place over medium heat and bring to a low simmer, stirring occasionally. Meanwhile, combine the pudding mix, sugar, and agar powder and gently mix to combine. Once the coconut milk is heated, slowly add the powdered mixture. Gently whisk together while keeping over medium heat until it begins to thicken, about 1 to 2 minutes.
3. Remove pan from heat and continue to whisk for 1 minute, allowing to cool a little. Add the lime juice and whisk together. Immediately pour into the cooled pie crust. Refrigerate for at least one hour to set.

You can add a little green food coloring if you want more of a green color.
You can use 1 1/2 cups of any crumbs of your choosing to help lower the protein or PHE.

Yields 8 servings
Serving size: 1 slice
1.6 g protein per serving
272 calories per serving

Fundraising Tip

Beer Bounce, Cocktail Crawl, Pub Crawl

So basketball isn't your thing, but an excuse to get a drink with your friends is? The Pub Crawl might just be the right thing for you! A pub crawl is an event in which a group of people visit multiple bars in one night. In order to raise money for an organization, gather as many participants as possible (that are legal drinking age) and start planning your course.

1. **Choose a date and starting time:**
2. **Choose your venues:**

- We recommend that you find an area that has multiple bars within walking distance of each other; this avoids the risk of anyone drinking and driving. Encourage everyone to take public transportation or use Uber/Lyft to get to and from the event.
- Once you get a path set up, contact the bar owners by email or stop in during the day when hours are slow to request a pub crawl. Their participation should include free entrance and designated pub crawl drink specials. Some venues may be willing to offer a free drink at their bar for participants in return of you putting their logo on the flyer.
- For bars that feature PA systems and a stage, ask if a volunteer may make an announcement when they arrive and announce they are raising money for HCU Network America.

3. **Create your flyer:**

- Include the price, date, time, route path (names of bars and their addresses), the times you will be at each venue, and what is included in the ticket.
- Include information about Homocystinuria or HCU Network America.

4. **Sell tickets in advance:**

- Range your ticket prices from $20-$40 depending on the size of your event.
- Ask the venues you approached if they would be willing to let you hang flyers
- Either use a ticketing service like Eventbrite, Ticketleap, RSVPify, or set up a PayPal account and email a receipt of their purchase.

5. **Let the fun begin:**

- On the night of the event, set up a booth or table at the first bar where participants can check in.
- Check ID's, take tickets, give wristbands. The wristbands will identify individuals as participants and grant them access to all of the pub crawl activities.

For more ways to host a fun and well attended pub crawl, visit: https://www.fundraiserhelp.com/pub-crawl.htm

Calling All Patients !

CALLING ALL PATIENTS WHO WERE MISSED BY NEWBORN SCREENING AT BIRTH!

WE HAVE AN OPPORTUNITY TO HELP CHANGE THE PROCESS BUT NEED YOUR STORY TO GIVE US THE EVIDENCE TO BUILD OUR CASE

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?

Later this year, a session on Newborn Screening for HCU will be held at a meeting of key leaders and staff from Newborn Screening Labs across the US. One of our medical advisors is preparing an abstract to help convince states to improve their process based on the large number of cases missed by the current approach. If you or your loved one were missed at screening, we need to hear from you ASAP so we have enough evidence to bring about change. Contact Danae if you can help us and she will lead you through the process that is outlined below.

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 vie-email 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

Texas HCU Family Picnic and OT-58

Registration now open: https://hcunetworkamerica.org/texas-family-picnic/

Orphan Technologies is enrolling patients into a first in human study of OT-58, an enzyme 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 five 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
- University of Miami – not yet open to 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. By registering, you will be able to identify other patients in your state and request their contact information. You will also 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 patients and families with resources, 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/

HCU Heroes

Rileigh

From Ohio

Our daughter Rileigh was diagnosed with Homocystinuria caused by Cystathionine Beta-Synthase Deficiency. Our story began in Ohio at home one and a half weeks after Rileigh was born. I got a phone call from her pediatrician's office. They told me that her amino acid test came back abnormal and that they needed to retest it. This is a part of the newborn screening program in Ohio and West Virginia. My advice to any parent is do not look things up on the Internet before knowing the final result! I was a basket case! I went to our local hospital and had her blood drawn again. A few days later my pediatrician's office called and said her amino acid was still abnormal, so they set up an appointment for us to go to Nationwide Children's Hospital in Columbus to see a Geneticist. We were told that "we were in good hands and that I know you are scared but everything is going to be okay. Fortunately, this is a treatable condition and as long as you do everything we ask of you, she will be okay and not suffer from any of the symptoms that this condition can cause." Okay, "so breathe, stop crying... he said she was going to be okay....". Learning that your child has a condition can be the scariest thing you'll go through. Learning that she will be okay as long as we do everything right can also be scary. "What if I fail? What if I do everything right but it doesn't work?" Your mind can be a horrible place to live in sometimes....

We put our worry to the side and did what was necessary. She was B6 non-responsive, so she gets a daily dose of B6 and Folic Acid. She also takes betaine with her daily formula, and is limited in the amount of protein she gets daily. We are 19 months out from diagnosis. She is thriving and doing very well! We couldn't be happier, and yes I've learned to trust the process.

The process isn't easy though. Her diet has been easily controlled with methionine free formula and baby foods. We use the nutrition labels to watch her protein intake and follow the nutritionist's advice step-by-step. I've found gluten free snacks, and she can have most fruits and several veggies. She loves bananas and green beans the most! Right now she's allotted 9 grams of protein a day from foods. I've been transitioning her to more solid foods; learning her likes and dislikes. Meal prepping for her caregivers while I'm at work has been the most challenging, but they have been great at keeping her on the diet. I have written out what she is allowed to have daily to make things easy on them. I know I will be stepping into another challenge once she is in school, but for now I have found some wonderful support groups that share terrific recipes and advice until that day comes. Our family has embraced our new normal and we adore our sweet, beautiful baby girl Rileigh.

March Madness

March Madness is a three week period packed full of buzzer beaters and is a sports poolers dream. But how can you keep the excitement alive when most of your members have had their brackets busted? A great option is our Madness Squares pool format, as every game of the tournament will have a winner!

How Do Madness Squares Work?

If you are familiar with Super Bowl Squares, the main idea is the same for March Madness. A 10x10 grid of boxes is setup and each row and column is given a number from 0 to 9. Just like in Super Bowl Squares, each square of the grid can be claimed by a pool member.

Winner breakdown

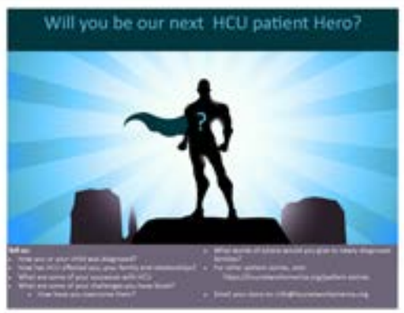
Each round is worth a set number of points, you can determine this on your own, but be sure to let all of the entries know before the tournament begins what the scoring system will be. (You should write the point values under each round at the top of the bracket).

Declaring a Winner

Multiply the total number of correctly picked games in each round by the points assigned to that particular round. Tally all rounds together and the person with the highest point total wins!

For further instructions and to print your bracket, visit:
https://www.printyourbrackets.com/howtomarchmadness.html

For online tools, check out:
https://www.runyourpool.com/march-madness-squares-pools.cfm



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

Focused on the Few™

At Recordati, we focus on the few - those affected by rare diseases. They are our top priority and at the core of everything we do. Our mission is to reduce the impact of extremely rare and devastating diseases by providing urgently needed therapies. We work side-by-side with rare disease communities to increase awareness, improve diagnosis and expand availability of treatments for people with rare diseases.

Recordati Rare Diseases is proud to support HCU Network America in their commitment to people living with HCU.

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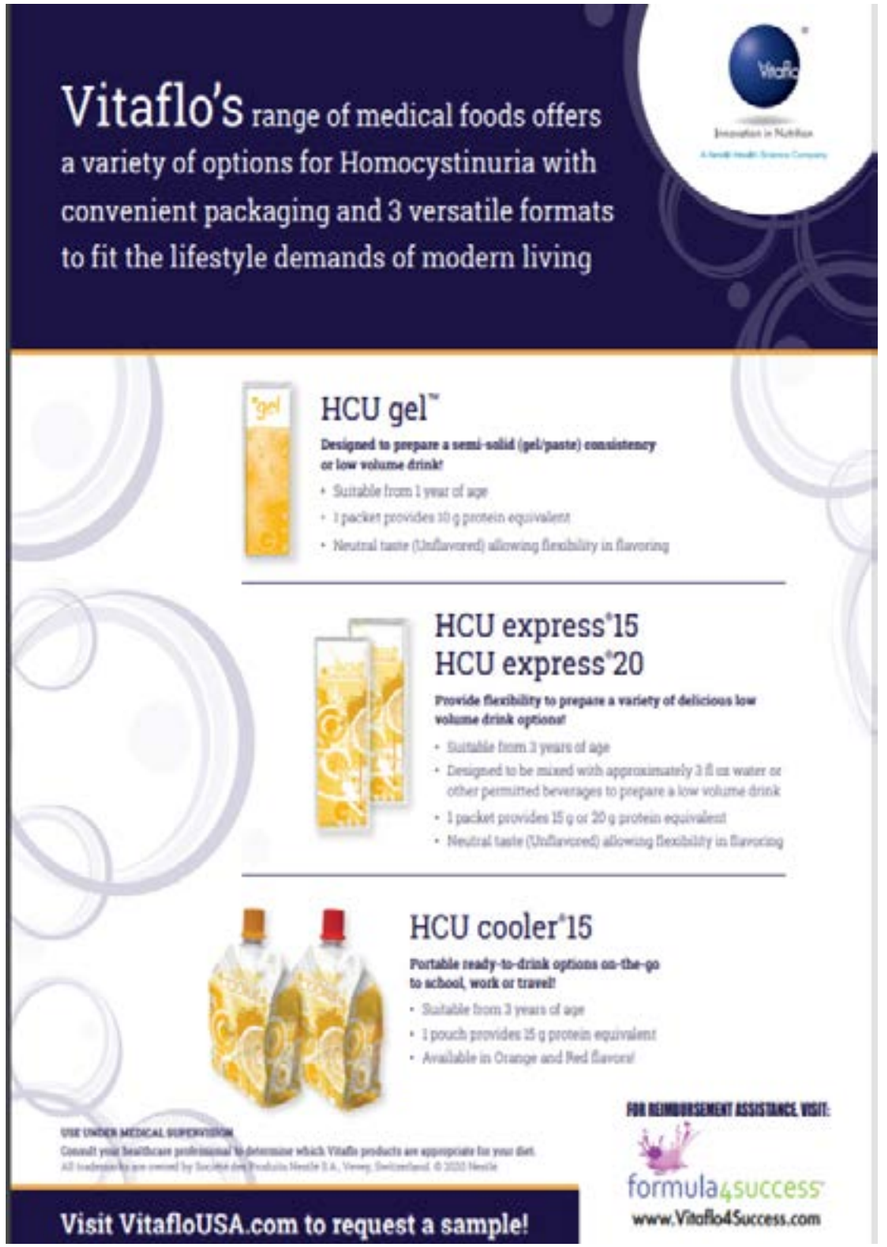

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