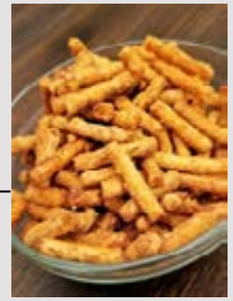
THE HCU Herald

Recipes from the Kitchen with Amber Gibson

Spiced Pretzels



Servings: 5.6
Serving size: 25 g
Protein per serving: 0.2 g
Calories per serving: 127

Love the smell of the roasted nuts you can get at the malls during the holiday season? You are in luck! Here is a low-protein version for all to enjoy!

Ingredients:

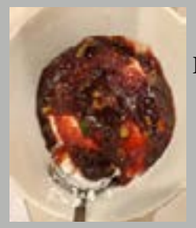
- 80 g Gluten-Free Pretzel Sticks, Broken in Half
- ½ c Sugar, White Granulated
- ¼ c Water
- ¼ tsp Vanilla Extract
- ¼ tsp Cinnamon, ground

Directions:

1. Add sugar and water to a medium skillet and place over medium heat. Stir with a wooden spoon until the sugar has completely dissolved. Bring to a simmer, then add the pretzels. Continue to stir while coming to a boil. Allow to cook, stirring constantly until the water evaporates and sugar begins to crystallize. It will take at least 5 to 10 minutes, so be patient and keep stirring.

2. Once sugar begins to crystallize, remove from heat and add the vanilla and cinnamon. Stir well and pour onto parchment paper and allow to cool completely. The pretzels will be crunchy once completely cool.

Thanksgiving Dip



Servings: 4.8
Serving Size: 2 Tbsp
Protein per serving: 0.3 g
Calories per serving: 69

While it has the name Thanksgiving in the title, this tasty dip is great for any fall/winter gathering!

Ingredients:

- 6 fl. oz. Cranberry Sauce, Jellied or Whole berry, canned ½ can
- 2 Tbsp Diced Jalapeno
- 1 Tbsp thinly Sliced Scallions
- ¼ tsp Salt
- ½ tsp Bragg's Coconut Aminos
- ¼ tsp Cambrooke Chicken-Flavored Consommé & Seasoning, Dry
- Optional: Cambrooke – Cream cheese (not included in the nutritional info)

Instructions:

1. Add all ingredients into a small bowl and mix well to combine. Pour into a glass jar with a lid and refrigerate 4 hours or up to overnight for flavors to meld together. Serve over Cambrooke's cream cheese for a nice creamy dip.

Note: You can use bell peppers in place of jalapeno peppers to pull down the spice a bit.

Edible Cookie Dough

Servings: 5.45

Serving Size: 62 g

Protein per serving: 0.7 g

Calories per serving: 260

Ingredients:

- 4 Tbsp. Butter, regular or unsalted, softened
- 6 TBSP Sugar, Brown, packed
- 25 g Vanilla Pudding, dry mix only
- 84 g Wheat Starch
- 3 TBSP Coffeemate, liquid, all flavors except Chocolate
- 50 g Semi-Sweet Chocolate Chips

Optional: If you prefer to make it a cookie dough dip – Fold in coco whip or cool whip and serve with low protein graham crackers (not included in nutritional info).

Directions:

1. Place softened butter and brown sugar in bowl of stand mixer with paddle attachment. Blend on medium speed until light and fluffy, about two minutes.

2. Add the pudding mix and wheat starch. Mix on low speed until dry ingredients are mixed with butter mixture. It will be crumbly. While on low speed, add the non-dairy creamer one tablespoon at a time until it begins to look like cookie dough batter

3. Turn off mixture and fold in chocolate chips. I recommend putting the cookie dough in a sealable container for at least 20 minutes to help harden the dough a little bit. Enjoy!

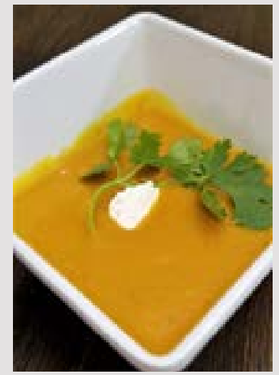
Butternut Squash Curry Soup

Servings: 6.19

Serving Size: 8 oz.

Protein per serving: 2.4 g per serving

Calories per serving: 199



Ingredients:

- 2 Tbsp Oil, Olive
- 65 g Diced Onion
- 52 g Diced Carrots
- 52 g Diced Celery
- 2 clove(s) Garlic, fresh cloves, chopped
- 1 tsp Grated ginger root or ginger paste
- 50 g Diced Bell Pepper
- 530 g Small Diced Butternut Squash
- 120 g Diced Parsnips
- 3 c Vegetable Broth
- 1 packet(s) G. Washingtons Golden Seasoning & Broth mix
- 13 fl.oz. Coconut Milk, canned
- 1 tsp Yellow Curry Powder

Instructions:

1. In a large saucepan over medium heat, heat the olive oil. Add the onions and saute until translucent, about one minute. Add the carrots, celery and garlic. Cook for about four to five minutes, stirring frequently to prevent burning. Add a tablespoon or two of olive oil if the vegetables look a little dry. Add the peppers and ginger and saute for one minute.

2. Add the butternut squash, parsnips, vegetable broth, seasoning packet, coconut milk, and curry powder. Stir to combine. Bring to a simmer and cook until the vegetables are tender, about 25 minutes. Using a blender or an immersion blender puree the soup until nice and creamy. Season with salt and pepper to taste. Serve hot.

Notes:

You can serve this soup with a garnish of cilantro, a small dollop of sour cream, and a lime wedge. Low protein rice would be a nice accompaniment to this dish!

Support H.R. 2507/ S. 2158, the Newborn Screening Saves Lives Reauthorization Act

Legislation to reauthorize the Newborn Screening Saves Lives Act was introduced in the House of Representatives by Representative Lucille Roybal-Allard on May 2, 2019 and in the Senate by Senator Maggie Hassan on July 18, 2019. H.R. 2507/ S. 2158, the Newborn Screening Saves Lives Reauthorization Act, will continue critical federal programs that provide assistance to states to improve and expand their newborn screening programs, support parent and provider education, and ensure laboratory quality and surveillance for newborn screening. Without reauthorization, these programs will expire at the end of Fiscal Year 2019

As an advocate for patients with rare diseases you are a very important part of the legislative process. You can make the difference as you are the voices your legislators want, or in some cases do not want, but need to hear. Please click here to complete the online form to take action and contact your Members of Congress. This bill has been stalled in the Senate due to an amendment introduced that would stifle research – we need you to help us overcome this block.

After you take action your job is not done! The final step is to share the action alert with your family, friends, co-workers and any other people that might be interested in taking action on behalf of the rare disease community

Alternatively you can call 1-844-872-0234 and wait for the automated voice machine. Press "1" and enter your ZIP code XXXXX. This will connect you to your first senator.

Ask: Please cosponsor the Newborn Screening Saves Lives Reauthorization Act

Hi! My name is XXXX and I'm calling from [your city/town].

As a [patient or caregiver] in the rare disease community, I ask that you please cosponsor H.R. 2507, the Newborn Screening Saves Lives Reauthorization Act without the harmful informed consent amendment. This legislation will reauthorize critical federal programs that provide assistance to states to improve and expand their newborn screening programs, support parent and provider education, and ensure laboratory quality and surveillance for newborn screening. Without reauthorization, these programs will expire at the end of Fiscal Year 2019.

Newborn screening detects conditions that, if left untreated, can cause disabilities, developmental delays, illness or even death. If diagnosed early, many of these disorders can be managed successfullyand at a lower long-term cost. These public health programs were last reauthorized in 2014 with unanimous consent in both the House and Senate. Please stand with the 4 million babies born in the United States each year and cosponsor this legislation.

Please contact me at [your phone number] or [your email address] to let me know if you will support this effort.

Thank you for your service and for considering my request.

$20,000 Donor Match is Back!

Thanks to an anonymous donor, any **funds** you help raise from November through December 31, 2019 **will be matched up to $20,000!**

During the winter holiday's warm hearts and generosity can be felt near and wide. During this time, we ask that you share our appeal letter with your collegues, friends and family.

Made your donation?
Don't forget to submit it to your employer's Company Match program!

Rare Science

Are you interested in receiving a Rare Bear?

Rare Bear applications open in August of each year and then are gifted in October for HCU Awareness Month. If you'd like to get on the list for our next gifting, please reach out to Danae' Bartke, HCU Network America, Executive Director at dbartke@hcunetworkamerica.org

Looking for a unique holiday gift this winter ?

Check out our line of HCU Awareness shirts only at:
bonefire.com/store/hcu-haberdashery/

Be a HCU HERO !

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

HCU Heroes

Leo

From Florida

This is Leo, born September 2016 with Homocystinuria caused by a rare form of MTHFR Deficiency. He was born via c-section at 40 weeks and 1 day. My husband and I were so excited to meet our 4th child. Throughout my pregnancy I did have a feeling about something being different. It was a strange feeling but I couldn't put my finger on why, and it turned out my gut feeling was right. I was blessed with a rare little boy. Leo was born a healthy 7 pounds 6 ounces and had a precious first cry. Leo did have some skin issues, red bright patches on his face, noisy eating and wasn't very alert. He just slept and ate. This did not alarm the doctors because most newborns do sleep a lot. I still had that feeling in the pit of my stomach though. A few days had passed and we were sent home to start our new routine.

A week into Leo's arrival a phone call arrived. It was the hospital informing us Leo's newborn screening confirmed he had a metabolic disorder. He had extremely high levels of homocysteine. My stomach dropped and tears started coming down my face. I didn't know what to expect or even what a metabolic disorder was. The Gainesville, FL genetic team soon after met with me and told me Leo needed to have several blood and urine tests done to confirm his diagnosis. They had told me at first it was Homocystinuria so we tried learning everything we could about this disorder. He was 6 weeks old at that time and still the genetic team was waiting on approvals for medication and trying to figure out dosing for him. A week went by and I noticed Leo was lethargic and didn't feel that warm to the touch after being uncovered so I took him to his pediatrician. Once we arrived she did a rectal temp which turned out normal. He was alert then so we went home. Everything seemed okay.

Another week went by; Leo is now 2 months old. The morning of December 3, 2016 I noticed Leo sleeping through his feeding so I would wake him trying to get him to eat. He would not wake to eat and was back to feeling cooler than he should. He wasn't cold but his head was slightly cool. We got alarmed and took him to the hospital. Once we arrived the nurse confirmed Leo was 94 degrees, and I told the doctors what his condition was so they could call his genetic doctors in Gainesville. They never heard of HCU and were trying to read about it at the hospital. They did a spinal tap, checked his brain activity (which showed no activity), did blood and urine tests, x-rays, and a CT scan. We were there all day. By 11 pm they decided to have Shands Children's Hospital come pick Leo up by helicopter. We were told to go home and go to sleep and drive there the next day because it was almost midnight by the time Shands had picked him up. There was no way we could do that. We hit the road and it took us 5.5 hours to get there. At Six am the next morning we finally arrived. They had already done an MRI, finding a clot on the brain called Cerebral Venous Sinus Thrombosis.

Leo

From Florida

Leo was in the ICU for a little over a week. He had to have a heater, feeding tube and oxygen. Doctors started Cystadane and Leucovorin and a B12 shot along with Lovenox hoping that would help him. He slowly but surely started waking up and being active after days on all the medications, and it was the first time I had ever seen him this alert. It was so heartwarming to see him coming back to me. He slowly got off the heater followed by the feeding tube and oxygen. He made progress every day. He was released to us on December 16th and the doctors taught us how to administer shots as we would be continuing the blood thinners at home for 3 months. In March 2018 a new MRI showed the blood clot had disappeared!

A new test showed that Leo has a rare form of MTHFR Deficiency that causes Homocystinuria. This condition mocks the symptoms of classic Homocystinuria but instead Leo has high homocysteine and low methionine and does not need a strict diet. He has two copies of a common variant c.655>T and two more unknown variants that have little to no research, MTHFR c.773C>G and MTHFR c.168G>A. He is being treated with Cystadane, Leucovorin and Deplin. This treatment has been working well to lower his homocysteine and raise his methionine.

Leo has been receiving speech therapy, OT and soon PT. He has low muscle tone, microcephaly, developmental delays, some behavioral issues, very short attention span, processing issues, neurological issues, and farsightedness. Leo is very bright; we have discovered that he learns very well by music and singing, which we have done since he was a baby. He can count to 10 and sings his ABC's. He will soon be in an academy that is designed for children with delays which will help him even further! Leo loves books and really enjoys Spiderman right now. He is full of energy and loves to climb on EVERYTHING! He has always had a high pain tolerance as well. He is a very strong and free boy! I always say we are learning more about Leo every day and where he is going. We want him to live life just like any other child. Mommy and daddy will always be there to guide and advocate for him.

Open Enrollment

OPEN ENROLLMENT
Healthcare.gov premiums are dropping 4% for 2020 plans.

A note from Raenette Franco –
Certified Biller Coder Specialist/Consultant
Compassion Works Medical Food Reimbursement Specialists

Healthcare.gov premiums are dropping and are now more affordable. Also, open enrollment has already started for some health plans, but the actual start date for all plans is November 1, 2019 to December 15, 2019. Please make sure you and your family members or patients are aware.

There are a lot of different health plans and depending on which one you choose it could mean a whole lot for affordability and coverage for things such as premiums, co-insurance/ co-pays and deductibles.

Keep in mind, medical foods/formula and solid low protein foods are typically under the Durable Medical Equipment (DME) medical benefits. So, when choosing a plan for yourself or your family member or patient that requires medical foods, search for the summary of DME benefits.

Next, once a health plan is chosen, go to their website and search for any policies in place for medical foods, enteral nutrition, and nutrition to get a better idea as to how the health plan covers medical foods (especially orally). Once you confirm coverage and affordability then choose the health plan. All plans purchased under the Healthcare.gov or directly from any health plan website are considered “fully-insured health plans”. All state mandates apply to any exclusions (if your state has a mandate).

As always, if you have any questions or need coverage support for yourself or your patients feel free to contact me. The service is free!

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(973) 832-4736

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/