The HCU *Herald*

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HCU HERO: LYDIA FROM MINNESOTA

My husband AJ and I welcomed our first child, Lydia, on Christmas Eve 2017. Though she was five weeks early, she weighed a healthy six pounds even. When she was born, we heard everything from “she’s the best present” to “at least you get a tax break!” But, little did people realize, this also meant that we had to meet our out-of-pocket max for health insurance. Which we did twice unfortunately, and very quickly.

Due to her prematurity, Lydia was immediately brought to the Special Care Nursery where we stayed for 18 days. Luckily, she was breathing on her own and maintaining her own temperature. She just needed to learn to eat. Being first time parents, it was hard to adjust to caring for a tiny newborn who had cords all over her, while being confined to holding her in a 5 foot radius with non-stop beeping, and nurses coming and going at all times of the day and night. At the same time, we were lucky to get all the additional training and support from the nurses (who are real-life angels) and the close monitoring that we didn’t know she needed at the time. Anyone who’s had a newborn with an extended hospital stay understands the emotions that come with it; the pumping, the cafeteria food, the homesickness, the uncertainty. All you want is to go home and start your life as a new family. Some days I still wish that was the only trauma we had to deal with at the beginning of Lydia’s life.

Around day 3 or 4, we were told that Lydia had an abnormal result on her newborn screening and they would be running additional lab work. All we heard was, “don’t worry, we get false positives all the time,” and, “don’t worry, these diseases are so rare, it’s highly unlikely it’s actually anything.” We immediately started asking questions about what this could mean for Lydia but the nurse practitioner and neonatologist simply refused to engage us in anything related to it because it was “probably nothing.”

The first set of labs came back “borderline abnormal.” Not bad enough to make the doctors concerned, but bad enough to warrant another set of labs. Again they said, “it’s probably nothing,” “it’s probably just faulty results due to prematurity,” and, “her body’s probably still just trying to work itself out.” They decided to start treating her with Levocarnitine as a precaution. I wasn’t terribly concerned at this point because when you have enough professionals assuring you it’s nothing, you believe it. I was also so distracted with baby blues, which were intensified by being stuck in the hospital, that I really didn’t have the energy to worry. My husband AJ, on the other hand, was concerned and was doing his own research as well as bombarding the doctors with, “have the new labs come back yet?”

With Lydia’s birth and hospital stay being over the holidays, lab results were prolonged. By day 15, we still hadn’t gotten her third set of labs back and Lydia was drinking her required amount from a bottle! She had been feeding-tube free for 24 hours and we were thinking "oh my goodness, we could go home any day now"! A new neonatalogist that we hadn’t met yet came in bright and early and I was anxious with anticipation of “you get to go home today!”, when she started rambling about how “the results came back and are still abnormal and these results are not typical results seen in prematurity and I think she has ‘it.’” It took me completely off-guard. She said she was already trying to get in touch with the geneticist to see what they wanted to do. After asking, “but what is ‘IT’?” the doctor calmly said “MMA.” At that time, it was the closest we had gotten to any information related to what may be going on with Lydia. The doctor should have said “don’t Google it” (we would have anyway) because one of the first things I read about MMA was “people with MMA rarely live into adulthood.” This day went from “we get to go home!” to “what? Something very serious is actually wrong with my baby?” Ouch.

The neonatologist came back a while later and said, “the geneticist wants to transfer you to Children’s in Minneapolis so an ambulance will be coming in a couple of hours to bring you there.” This day will go down in history as one of the worst days of my life. Watching your seemingly perfectly healthy newborn put in an incubator and wheeled onto an ambulance being brought somewhere to learn she may not be perfectly healthy was a nightmare.

Soon after, the frequent appointments began. We met with geneticists and a genetic counselor who started genetic testing. Lydia’s two abnormal elevated levels were Methylmalonic Acid (MMA) and Homocysteine (HCU). Because of this, the geneticists suspected a Cobalamin deficiency disorder and wanted to start her on Cobalamin (B12) injections immediately. On day 18 we were able to go home after learning how to give our newborn shots (that will continue daily and for the rest of her life). We were sent home with 1 daily injection, 3 oral meds, medical formula, and directions to monitor her closely for signs of metabolic crisis such as lethargy, decreased eating, and vomiting (cue: extreme anxiety every time she vomits for the rest of her life).

After the first round of genetic testing came back normal, they dug deeper into the “rarer of the rare” disorders. On February 23rd, 2018, we received the phone call we were anxiously awaiting and also quite a bit dreading. Lydia was diagnosed with Cobalamin F disease, an extremely rare Cobalamin deficiency disorder. At the time of diagnosis, we were told there were less than 20 documented cases in the medical literature in the WORLD.

Cobalamin F is an autosomal recessive disorder (AJ and I are both carriers) and is a disorder of transport which means, put very simply, the B12 isn't able to get where it needs to go in her body. Once it's there (and we get it 'there' with injections), her body knows what to do with it so she is considered "B12 responsive," which we are thankful for. The injections have managed to bring her HCU and MMA into the normal range. When these toxins are not managed, it can lead to all sorts of terrifying things such as seizures, strokes, liver damage, intellectual disabilities, etc. Getting that phone call was hard. I always had it in the back of my head that maybe this was all just a huge fluke. Maybe her premature body took a while to start functioning and she didn't have anything at all. But genes don't lie and they found proof that something was wrong. There was no denying it now.

Our new normal began immediately after that phone call. We had to learn words like "homocysteine, methylmalonic acid, lysosome, cobalamin, pyridoxine, intramuscular and subcutaneous, and metabolic disorder." In addition, frequent medical appointments and an ever growing list of professionals joined our team including neurology, ophthalmology, cardiology, developmental pediatrician, Early Intervention, and eventually private speech and physical therapies.

Fast forward two years and some months later and Lydia is a thriving toddler! She tolerates the injections better than any two year old should have to. She has some developmental delays that are addressed through Early Intervention and outside therapies. She has hypotonia and motor planning difficulties. (Editors note: motor planning is a skill involved with remembering and performing steps to make a movement happen.) She's also downright perfect!

It's not easy having a child with a rare disorder and we find ourselves questioning whether every little thing is related to her disorder ("Oh my gosh, she stubbed her toe! Is this related to her CblF?"). We also find ourselves questioning whether she's getting the best treatment, which is so hard to know due to the rarity and, therefore, limited research about these disorders. She feels like our own little science project.

Throughout this process, we have made connections with other families that have come before us with metabolic disorders and we're grateful for their guidance because they've helped us get where we are today.

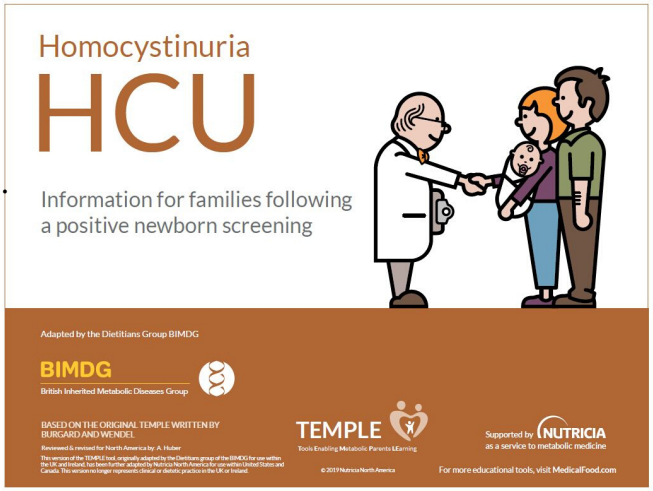
As time progresses, we plan to participate in studies to help contribute to science as a way to give back to the community that has given so much to us. Without newborn screenings being as advanced as they are today, there's no telling what our lives would look like. And for that, we will always be thankful.

NEWS YOU SHOULD KNOW

TEMPLE
Tools Enabling Metabolic Parents LEarning

TEMPLE makes learning about HCU easier for families and friends

TEMPLE (Tools Enabling Metabolic Parents Learning) is a leading education series for inborn errors of metabolism, including Homocystinuria (HCU). These booklets and videos explain the condition and its management in easy-to-understand language and pictures. They are ideal for educating new parents and families after a positive newborn screening. TEMPLE booklets and videos can also be used to teach grandparents, relatives, and friends about the condition. TEMPLE was created by Nutricia



Share TEMPLE with relatives and friends in your life to help them better understand the basics of your child's or loved one's condition. You can aind TEMPLE booklets on Nutricia's website and TEMPLE videos on Nutricia's YouTube channel

Exciting news from Nutricia is that they recently released TEMPLE booklets for HCU in Spanish and French! Pass along to those you know who may have Spanish- or French-speaking relatives or friends who would like to better understand the condition.

Learn more about Nutricia North America's products and services at MedicalFood.com

HCU Network America is aware of the social media posts circulating about HCU and COVID-19. Please see the below information from Dr. Kimberly Chapman, our medical advisor and attending physician in Genetics and Metabolism at Children's National Hospital, DC, and Assistant Professor of Pediatric and Integrated Systems of Biology at George Washington University in Washington, DC.

COVID-19 Statement

Thank you for your question about COVID-19 infection and risks for patients with Homocystinurias. I do not anticipate that individuals with Homocystinurias will suffer any greater risk of being infected with COVID-19 virus than anyone else. If you were to be infected then your complication risk and symptoms will probably be similar to the general population.

The Centers for Disease Control (CDC) has published guidance stating that individuals with inborn errors of metabolism might be at greater risk from COVID-19 than other individuals. The reason for this and homocystinuria is that your homocysteine is likely to increase during an infection. The best defense for this is to remain hydrated, continue your prescribed medications and diet (if prescribed).

COVID-19 is a serious disease. Everyone should obey whatever restrictions and guidelines are being recommended where they live. The most important things are to wash your hands frequently and if ill, particularly with fever, cough, vomiting, or diarrhea, isolate oneself at home and contact your primary care physician (and your metabolic providers) about how to be evaluated clinically, change your regimen and potentially tested for COVID-19. If you're feeling more severely ill, such as shortness of breath, then contact 911.

The most important thing to do with regards to homocystinurias and the system disruptions that are likely to occur in the next few weeks is to make certain that you have ordered and obtained at least one month's supply of medical food or medication.

A short list of things you can do to prepare for COVID-19 as well as generally for any mass infection or disaster.

1. Appropriate hand washing is essential. Practice washing your hands with your child using soap and scrubbing for at least 20 seconds (take this time to sing the ABCs out loud together). Here is a link to the CDCs information on hand-washing: https://www.cdc.gov/handwashing/when-how-handwashing.html

2. Try to have at least 1-2 weeks' worth of specialty medications and formula on hand. Even if something does not affect you in your immediate area, it is good to be prepared for delays in shipments.

Of note, insurance may not allow for an on-hand stock of medication, in this case, work with your metabolic clinic to develop an emergency plan for medication stocks.

For those on a metabolic dietary formula, consider building a stock that can cover 1-2 weeks. This will help you from running out (because we often forget to order things regularly).

3. In the event of a disaster it is a good idea to have extra food/water/juice on hand if something were to happen and you are unable to get to a grocery store for one week.

4. Some things you should keep in your pantry.

A. Families with kids > 1 year of age can stock sugar and Gatorade. Powdered Gatorade (or equivalent) is preferred, as it can be reconstituted in bulk into a large volume as needed. However, pre-made Gatorade is ok too. (1 tablespoons of sugar into 20 oz of Gatorade makes a solution that is ~10% dextrose)

B. Families with kids < 1 year of age should stock Pedialyte instead for re-hydration, as Gatorade may result in an osmotic diarrhea

C. Families should also stock antipyretics, Tylenol preferred over ibuprofen, given the recent (low-evidence) concerns regarding NSAIDs in COVID

5. If you have planned travel outside of the US please be aware of travel warnings from the CDC. These are updated daily and can be found here: https://www.cdc.gov/coronavirus/2019-ncov/travelers/index.html

Homocystinuria (Classical, Cobalamin, MTHFR) Emergency Toolkit

In October 2018 , HCU Network America's Patient- Parent Advisory Committee published the first Homocystinuria Emergency Toolkit. With the addition of the Cobalamin Steering Committee to HCU Network America we are working tirelessly to make sure our resources not only cover classical homocystinuria, but also cobalamin disorders and severe MTHFR.

In addition to including information for cobalamin disorders and severe MTHFR we have added a Medical Emergency Letter (one for each disorder) template that you should send to your medical team to complete ASAP!

When you have HCU, being ready to handle difficult situations requires special planning, in addition to the typical things everyone needs to consider in the event of an emergency or natural disaster. Here are some key things to think about and discuss with your family today. Prepare yourself in advance!

Download the new Homocystinuria Emergency Toolkit:
https://hcunetworkamerica.org/toolkits-and-checklist/

EVENTS

All HCU Family Picnics and Meetups are currently on hold

Because of the unexpected health crisis surrounding COVID-19, HCU Network America is taking the necessary precautions and postponing all state and regional meetups until further notice. When the CDC advises it's safe to have social gatherings again, we will update the community with new dates and times. We appreciate your patience and understanding during this time.

Go The Extra Mile for HCU is Now Live! Register Now

Set with the participants convenience, geography and their pace in mind, in 2019, Go the Extra Mile for HCU raised more than $4,000 to contribute to help Homocystinuria patients. These races are convenient, fun and competitive way to support the efforts of HCU Network America (HCUNA). We sincerely appreciate your support for this event and HCUNA and hope you will join us for Go The Extra Mile for HCU 2020.

Registration is $25 per participant
Please note: Online registration closes at 11:59 p.m. (EST), May 3rd, 2020.

How does a virtual race work?

A virtual race is a race that can be walked, ran, or biked from any location you choose. You can participate on the road, on the trail, on the treadmill (or stationary bike), at the gym or on the track (or even at another race). You get to run your own race, at your own pace, and time it yourself. You do not have to complete the miles all at once, in one day, or even a week. You can use the entire month to complete the race.

Did you say "swag"?

All participants will receive a t-shirt, medal, and virtual bib.

More questions about the race?

Visit, https://hcunetworkamerica.org/virtual-race/

Covid 19-Impact

We hope to still be able to hold this event and stay consistent with the recommended social distancing directions in your area. This may require some individuals to do their miles "indoors". If we choose to postpone this event, we will let you know immediately.

RARE DISEASE DAY WITH CODEXIS INC

Across the globe, the last day of February each year is dedicated as Rare Disease Day. This year we had the honor of being invited to hang out with Codexis Inc at their company annual Rare Disease Day event. They day was designed to highlight not just Homocystinuria, but also Phenylketonuria (PKU) and Maple Syrup Urine Disorder (MSUD) – all inborn errors of the metabolism that Codexis is dedicated to.

The event started with a warm greeting and background on Rare Disease Day from their Sr. VP of Strategic Development, Gjalt Huisman. The day then included a combination of Clinician Presentations from Dr. Greg Enns, Natalia Gomez-Ospina and Dr. Kristina Cusmano-Ozog, along with panels featuring the clinicians, and a variety of patient and organization panels. The day ended with a tour of Codexis labs!

It was truly a great event to help spread awareness, not just for HCU, but for our other allied disorder families and rare disease as a whole! Thank you Codexis for the invitation and for the wonderful event

Left Picture: L to R: Diane, John and Mia Pytel (California Coalition for PKU and Allied Disorders, Jordann Coleman (MSUD Family Support), Danae Bartke (HCU Network America), Dr. Greg Enns (Stanford University), Dr. Natalia Gomez – Ospina (Stanford University), Dr. Kristina Cusmano-Ozog Stanford University)and Kevin Costa (SynBioBeta)

About Codexis

Codexis is a leading protein engineering company that applies its proprietary CodeEvolver® technology to develop proteins for a variety of applications, including as biocatalysts for the commercial manufacture of pharmaceuticals, fine chemicals and industrial enzymes, and enzymes as biotherapeutics and for use in molecular diagnostics. Codexis' proven technology enables improvements in protein performance. For therapeutic enzymes, this means to improve stability against the harsh environments it would encounter inside the body. Our goal is to develop novel therapeutics where a critical need exists, as well as improve efficacy for patients. Codexis is partnered with Nestle Health Science to advance CDX-6114, an oral enzyme therapeutic for Phenylketonuria, currently in Phase 1b.

For more information, see www.codexis.com

APRIL FUNDRAISING TIP: AMAZON SMILE

Not using Amazon Smile? It's easy, here's how!

What is Amazon Smile?
Amazon Smile is a simple and automatic way for you to support HCU Network America every time you shop, at no cost to you. When you shop, you'll find the exact same low prices, vast selection and convenient shopping experience as, with the added bonus that Amazon will donate a portion of the purchase price to us.

How do I set it up?
Simply, go to smile.amazon.com, the first time you go it will ask you to designate an organization. Type in HCU Network America and select us from the list (or go to our direct link: click here). It is important to note that in order for the donations to go to HCU Network America, you MUST check out from this url every time - see best practices below for some pointers on how to do this.

What if I'm already set up and would like to switch to HCU Network America?

1. From your desktop, simply select "Your Account" from the navigation at the top of any page
2. Then select the option to "Change your Charity". From your mobile browser, select "Change your Charity" from the options at the bottom of the page.
3. Type "HCU Network America" in the search bar and search for the charity.
4. Select HCU Network America charity to update your account

Best practices for using Amazon Smile
Now that your account is set up to use Amazon Smile, it is important to note that Amazon only makes donations to HCU Network America when you checkout from your cart from this url. This is the only way HCU Network America gets any donations from Amazon Smile. Since this is the case here are some best practices to help you make the most of your Amazon Shopping.

Desktop Users:
If you do your Amazon shopping from you desktop/laptop then you can simply bookmark/favorite this url and do your shopping from this web page.

Mobile Users:
Most Amazon shoppers use the app on their mobile or tablet. If you are an Android user, make sure to download the Amazon Smile App. All iPhone users, continue to do your shopping with the app then the only thing you have to change is how you do your checkout when you are done shopping. You will want to have this url saved as a favorite on your web browser (iPhone – Safari). When you are ready to do your checkout in Amazon, make sure your cart is loaded with your items, then simply exit the app and go to the this bookmarked page to continue your checkout.

RECIPES FROM THE KITCHEN:

Apple Fritters

Makes 12 servings

Ingredients:

- 1 c CFL Pancake and Waffle Mix
- 2 TBSP Sugar, Brown, packed
- 1 tsp Cinnamon, ground
- 1/4 tsp Allspice, ground
- 1/2 tsp Salt, Table
- 80 g Rice Dream, Original
- 1 tsp Vanilla Extract
- 80 g Diced apples, one medium apple

Directions:

1. Heat 1 qt vegetable oil in a saucepan over medium heat until temp reaches 375 degrees.

2. Mix the pancake mix, brown sugar, cinnamon, allspice, and salt in a medium bowl. Add the rice milk and vanilla and whisk to combine. Fold in the diced apples. Drop spoonful into the hot oil and fry until golden brown. Serve hot with icing, powdered sugar, syrup, or cinnamon and sugar

Nutritional Information

- Serving size: 1 Fritter
- Protein per serving: 0.2 g
- Calories per serving: 60

RECIPES FROM THE KITCHEN:

Banana Muffins

Makes 12 servings

Ingredients:

- 204g CFL baking mix
- 1 tsp cinnamon
- 1/2 tsp ginger
- 1 tsp baking powder
- 1 tsp baking soda
- 1/2 tsp salt

- 280g bananas, mashed
- 1/3 cup Sour Cream or Coconut Yogurt
- 1 tsp vanilla
- 1/2 cup brown sugar
- 3 Tbsp (36g) aquafaba, liquid from canned chickpeas
- 12 Tbsp butter, melted

Nutritional Information

- Serving size: 1 Muffin
- Protein per serving: 0.71 g

Directions:

1. Preheat oven to 350 degrees Fahrenheit. Line a muffin tin with cupcake liners.

2. Combine dry ingredients in a small bowl and lightly whisk to combine. Set aside.

3. In a bowl of a stand mixer, add the mashed bananas, sour cream, and vanilla. Mix to blend them together. Add the brown sugar and mix just until combined. Then add the melted butter and aquafaba. Mix until just blended. Don't over mix.

4. Add half the dry mixture and mix on low speed until dry ingredients are blended with the wet ingredients. Now add the remaining dry ingredients and mix just until blended.

5. Immediately scoop batter into prepared muffin, dividing evenly into the 12 liners. Bake in preheated oven for 10-12 minutes until a toothpick inserted into the muffins comes out clean.

THANK YOU, CHEF AMBER GIBSON

RECIPES FROM THE KITCHEN:

Cookie Butter Swirled Mini Banana Bread Loaves

Makes 9 servings

Ingredients:

- 2 fl.oz. Rice milk
- 1/2 tsp Lemon Juice
- 190 g Cambrooke Baking Mix, Lightly packed
- 2 tsp Baking Powder
- 3/4 tsp Salt, Table
- 1 tsp Cinnamon, ground
- 1/4 tsp Nutmeg, ground
- 8 TBSP Butter, regular or unsalted, softened
- 1/2 c Sugar, Brown, packed
- 200 g Banana, fresh, peeled, medium, mashed
- 1 tsp Vanilla Extract
- 6 g Unsweetened Vanilla Cultured Yogurt Alternative, about 2 Tablespoons
- 2 TBSP Biscoff Cookie Butter

Directions:

1. Preheat the oven to 350 degrees. Spray a mini loaf pan with nonstick cooking spray and set aside. In a small measuring cup combine the rice milk and lemon juice and allow to sit a few minutes to sour.
2. Combine the baking mix, baking powder, salt, cinnamon, and nutmeg in a medium bowl and lightly whisk to combine. Set aside.
3. In the bowl of a stand mixer add the softened butter and brown sugar. Cream together until light and fluffy. Add the vanilla, the mashed bananas, and the coconut milk yogurt. Mix just until combined. Add the dry ingredients to the wet ingredients and mix until combined. Add the sour rice milk and mix briefly to incorporate.
4. Fill the cavities of the mini loaf pan about half way with the batter. Put about 1/4 tsp of the cookie butter in each batter cavity and swirl with a tooth pick. Bake at 350 for 25 to 30 mins, until lightly browned and a toothpick inserted in the middle comes out clean. Remove from oven and allow to cool about 5 to 10 minutes then move to a wire rack to finish cooling.

Nutritional Information

- Serving size: 1 loaf
- Protein per serving: 0.5 g
- Calories per serving: 270

THANK YOU, CHEF AMBER GIBSON

RECIPES FROM THE KITCHEN:

Pot Pie Pasta

Makes 3 servings

Ingredients:

- 80 g Penne, dry (Low Protein)
- 100 g Jackfruit, Young in Brine, drained and seed removed, roughly chopped
- 1/2 tsp Italian Seasoning
- 2 TBSP Oil, Olive, divided
- 60 g Mushrooms, white or brown (Cremini), medium, whole, diced
- 30 g Diced Onions
- 1/2 c Thinly Sliced Carrots
- 1 clove(s) Garlic, fresh cloves, chopped
- 1/2 c Vegetable Broth
- 1 tsp Chicken-Flavored Consommé & Seasoning,
- 1/2 c Coconut Milk Beverage, Unsweetened
- 1/8 tsp Curry Powder

Directions:

1. Cook the penne pasta, rinse, and set aside.
2. Place the chopped jackfruit in a bowl and toss with the Italian seasoning. Set aside. In a medium skillet, preheat 1 tablespoon olive oil over medium heat. Add mushrooms and sauté until lightly browned. Remove from pan and set aside.
3. Heat one tablespoon oil over medium heat. Add the onions and carrots and sauté for 5 minutes. Add the garlic and sauté for one minute, stirring constantly. Add the jackfruit and sauté for 3 to 4 minutes. Add the vegetable broth, consommé powder, coconut milk, and curry powder. Cook on medium heat until thickened and carrots are fork tender. Add the cooked penne and gently toss. Serve immediately.

Notes: You can use any nondairy milk you choose. Just adjust where necessary.

Nutritional Information

- Serving size: 1/2 cup
- Protein per serving: 1.5 g
- Calories per serving: 196

RECIPES FROM THE KITCHEN:

Triple Ginger Cake

Makes 9 servings

Ingredients:

- 320 g MixQuick Baking Mix, gently packed, about 2 cups
- 1 1/2 tsp Ginger, ground
- 1/2 tsp Cinnamon, ground
- 1/2 tsp Salt, Table
- 1 tsp Baking Powder

Wet Ingredients

- 8 TBSP Butter, regular or unsalted, softened
- 1/2 c Sugar,Brown, packed
- 2 banana(s), fresh, peeled, medium, mashed
- 2 TBSP Mayonnaise
- 1 tsp Minced Fresh Ginger Root
- 1/4 c Chopped Candied Ginger
- 1/2 c Coffeemate Nondairy Creamer

Nutritional Information

- Serving size: 1 slice
- Protein per serving: 0.8 g
- Calories per serving: 371

Directions:

1. Preheat oven to 350 degrees. Spray an 8x8 pan with cooking spray and set aside.
2. Combine the dry ingredients in a medium bowl and gently whisk to combine. Set aside.
3. In the bowl of a stand mixer with the paddle attachment, cream the butter and brown sugar until light and fluffy. Add the bananas and mix just until combined. Now add the mayo and mix for 30 seconds. Add the fresh ginger and the candied ginger. Mix for another 30 seconds. Now add 1/3 of the dry ingredients and mix just until combined. Add 1/4 cup of the nondairy creamer and mix just until combined. Repeat until all ingredients are mixed, ending with the dry ingredients. Pour into prepared 8x8 pan. Bake for 45 to 50 minutes, or until a toothpick inserted in center comes out clean.

THANK YOU, CHEF AMBER GIBSON

MEET OUR NEWEST PATIENT-PARENT ADVISORY COMMITTEE MEMBER

Ruby is a working mother of a wonderful, rambunctious and high-spirited little boy. She and her husband, Stephen, learned about Homocystinuria when their son was diagnosed during her 2nd trimefster. Mount Sinai conducted a comprehensive test of genetic conditions on Ruby during her pregnancy and as a result of returning a positive result for HCU, they tested her husband. When her husband also returned a positive result for HCU (and the chances of their child being affected with HCU went from 1 in 200,000 to 1 in 4), Mount Sinai performed a CVS test on their unborn son. Having the diagnosis pre-birth really gave Ruby and Stephen a head start on preparing for what this meant for them and their child and began arming themselves with information and the tools they needed for their son prior to his birth. During this journey it soon became apparent that Homocystinuria is not a well-known genetic disorder among the medical field. A lot of guidance and information they obtained came from other parents across support groups online. Ruby hopes to be able to share her experience with other families and be able to support them and answer any questions they may have pertaining to maneuvering and managing this condition. To be able to contribute and provide the same support that her family initially received, and continue to receive, is something she strives towards as well as helping in raising awareness of a genetic condition that is not widely well known.

Our mission:

The Patient-Parent Advisory committee will come together to provide advice on how best to support the needs of the HCU community.

Learn more about Patient Parent Advisory Committee and it's other members at: https://hcunetworkamerica.org/patient-parent-advisory-committee/

Goals/Responsibilities:

1. Provide ideas to HCUNA on what would be helpful to the HCU community, e.g. through periodic calls with the executive director and other board members as needed
2. Act as an ambassador to new HCU families - providing them with information, tools, and advice
3. Review draft content and materials for new resources being developed by HCUNA and provide feedback
4. Help serve as or organize volunteers as needed for events such as the next Patient-Expert meeting (registration, t-shirts, goody bags, etc)

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