The HCU *Herald*

Newborn Screening and Family Planning Edition

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HCU HERO:
RAIDIN FROM IRAN

I was very upset when my second son showed the first symptoms of the disease at the age of 7 months. I lost my first son at the age of 6 months after enduring a month of hospitalization and not being diagnosed with the disease. I was scared to repeat! I cried for months, and I was not sure if I should to take my son to the doctor because they did not have
enough knowledge and information.

The symptoms were getting worse in my son and something inside me was screaming, “go to a doctor.” Maybe it’s just a simple vitamin deficiency. At 12 months, the symptoms turned into seizures, my son had spasms lasting a few seconds. His mental and physical development had stopped at 8 months.

When I first took him to a pediatric neurologist, my son was 17 months old. The doctor started treatment with anticonvulsants. I asked the doctor to prescribe a genetic test for my son. It was important for me to understand why my children were getting sick. The doctor told us that genetic testing has no effect on the treatment process and is very expensive.

A month after starting treatment, I met a friend who convinced me to have a genetic test as soon as possible. We did the testing when my son was 20 months old, but the doctor still believed that the test results would not change the treatment. Six months later, when my son was 26 months old, the genetic test results were ready, he wrote: MMACHC! (Editors note: this is the gene that causes Methylmalonic Aciduria with Homocystinuria Type C, also known as Cobalamin C.)

I started researching about this disease. I did not find answers to my questions in Persian sources, so I researched in English sources. I was a member of Facebook, so I looked for a group associated with this disease. After becoming a member of the MMA family group, I realized that the treatment of Iranian doctors for my son's disease was old and outdated. Doctors in Iran did not accept daily injections of hydroxycobalamin for my son's disease and told us to take oral medication. My friends on my Facebook group guided me. They informed me about the latest treatment methods, and I was able to find a doctor in Iran who accepted these methods and followed my son's treatment.

As I write this, my son is 3 and a half years old. We have come a long way in diagnosing and treating him. We are still battling seizures, but we are very close to victory! There were two rescue angels on my son's journey. The first angel was the one who convinced me to have a genetic test as soon as possible (Zari dear). The second angel was dear Noras Pasko, who found out I had to communicate with the members of the Facebook group in English, which I am not fluent in, so she explained everything to me in Persian very patiently.

Currently, I help families with MMACHC patients in Iran to get accurate and up-to-date information about the disease and treatment, although many of them believe that the doctor's instructions are better for them than the information of a housewife! Unfortunately, the injection of hydroxycobalamin is still ignored among Iranian doctors, and they prescribe the oral type of vitamin B12, which is sometimes cyanocobalamin.

MEET OUR NEWEST MEDICAL ADVISOR: JANET THOMAS

Professor, University of Colorado School of Medicine, Department of Pediatrics, Section of Clinical Genetics and Metabolism. Board certified Clinical Genetics and Clinical Biochemical Genetics.

Born and raised in Iowa, Dr. Thomas attended Loras College in Dubuque, Iowa for an undergraduate degree in biology before attending the University of Iowa College of Medicine. She did her pediatric training at the University of Arizona Health Sciences Center and then completed her genetics training at the University of Colorado School of Medicine. Subsequently, she joined the faculty of the University of Colorado School of Medicine in 1996 and has since remained there as an active member of the department of Pediatrics, Section of Clinical Genetics and Metabolism.

Dr. Thomas has devoted most of her time to caring for individuals of all ages with inborn errors of metabolism in the Rocky Mountain region. She has a particular interest in lysosomal storage disorders (especially the MPS disorders), phenylketonuria, newborn screening, and regional care. She is a faculty member of the UCDHSC Human Medical Genetics Program and a member of the State of Colorado Newborn Screening Program Stakeholders Committee. In addition, she is the Co-Program Director for the Mountain States Regional Genetics Network, an organization focused on regional and national delivery of genetic services including underserved populations, telegenetics, consumer advocacy and resources, and health care policy. Past positions have included Interim Section Head of the Section of Clinical Genetics and Metabolism, the Director of the Inherited Metabolic Diseases Clinic at Children's Hospital Colorado, and Director of the University of Colorado Medical Genetics Residency Program. Teaching medical students, pediatric and genetic residents, and graduate genetic counseling students is also a part of her duties. She has been actively involved in therapeutic research for PKU, lysosomal storage disorders, urea cycle disorders, and homocystinuria.

HCU COMMUNITY COOK BOOK

Sweet Potato, Carrot, and Ginger Soup

By Amber Gibson

Makes 6.6 servings

Ingredients:

- 2 TBSP Olive Oil
- 1/3 c Diced Raw Onions
- 2 carrot(s) Raw Carrots, peeled and diced
- 3 clove(s) Garlic, minced
- 1 TBSP Minced Ginger Root
- 2 TBSP Mirin Sweet Rice Seasoning
- 1 TBSP Rice Vinegar
- 305 g Peeled and Large Diced Sweet Potatoes
- 3 1/2 c Vegetable Broth
- 1 TBSP Cambrooke Chicken-Flavored Consommé & Seasoning, dry
- 3 TBSP Bragg's Coconut Aminos
- 1 tsp Orange Zest
- 1/4 c Orange Juice

Nutritional Information

- Serving size: 6 oz
- Protein per serving: 1.5 g
- Calories per serving: 120

Directions:

1. In a large saucepan, add the olive oil and heat over medium heat. Once the oil is ready, add the onions. Sauté until translucent, about 2 minutes. Add the diced carrots and sauté for three minutes. Now add the garlic and ginger and sauté for another two minutes. Make sure to stir constantly to prevent burning the garlic and ginger.
2. Add the mirin, rice vinegar, and sweet potatoes. Stir and cook for another three minutes. Add the vegetable broth, consommé seasoning, and coconut aminos. Bring to a simmer and cook until the carrots and sweet potatoes are fork tender. This should take about 30 to 40 minutes.
3. Remove pan from heat. Using an immersion blender, standard blender, or food processor, puree the soup until smooth. Return soup to pan. Add the orange juice and orange zest. Stir well. Season with salt and pepper as needed. Serve immediately.

HCU COMMUNITY COOK BOOK

Jackfruit Cakes

By Amber Gibson

Makes 3 servings

Ingredients:

- 95 g Canned Jackfruit, rinse well, seeds removed
- 1/4 c Cambrooke Seafood Patty Mix
- 2 tsp Olive Oil
- 1/2 c Water
- 1/8 tsp Old Bay Seasoning

Sauce:

- 1 1/2 TBSP Cambrooke Cream Cheese
- 1/4 tsp Sriracha
- 1 tsp Lemon Juice
- 1 TBSP Sour Cream

Nutritional Information

- Serving size: 1 cake
- Protein per serving: 0.6 g
- Calories per serving: 84

Directions:

1. Squeeze excess liquid from jackfruit. Roughly chop the jackfruit. The small chunks will mimic the texture of crab meat. Add the rest of the ingredients for the cakes from the seafood mix to the Old Bay Seasoning. Mix with a spatula until combined. Add more water a tablespoon at a time if a little dry
2. Add 1/4 c vegetable oil to a small skillet and place over medium heat. Form cakes in your hand about 3 inches wide and 1 inch thick. Place in preheated skillet and cook until browned on both sides and heated through, about 5 minutes per side. Remove from skillet and place on a paper towel lined plate to absorb excess oil..
3. For the sauce, combine ingredients and mix well. Serve with the jackfruit cakes.

GET READY!
OCTOBER IS HCU AWARENESS MONTH

This year's theme for our virtual race and for HCU Awareness Month is Go the Extra Mile for HCU. How do you exactly go the extra mile for HCU? You raise awareness, you fundraise, you advocate and help amplify the messages of the HCU community.

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

HCU Awareness T-Shirt:
Get your official 2020 HCU Awareness T-Shirts. While you wear your shirt, you help raise awareness for HCU, and by purchasing you are helping raise funds to benefit HCU Network America's Educational Program Fund. So order your shirt now so you have it in time for HCU Awareness Month!

Click the link to purchase - https://bonfire.com/go-the-extra-mile-for-hcu-homocystinuria

Can't race? Help spread awareness through the HCU Click Campaign!

Get your mouse ready and be prepared to click! For one week only (September 21-25) Recordati Rare Diseases will donate $5 (up to $5,000) to HCU Network America for every click. Be on the lookout for the link – you can click the link once a day and be sure to share it each time!

GET READY!
OCTOBER IS HCU AWARENESS MONTH

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. Additionally, look out for our fact of the day

HCU Awareness Month Activity List

- ☐ Change your social media picture to the HCU Awareness Ribbon
- ☐ Start a HCU fundraiser
- ☐ Share an infographic about HCU
- ☐ Share a patient story
- ☐ Share your diagnosis story
- ☐ Challenge your friends to the same amount of protein and three normal protein shakes a day #ToastTocHCU
- ☐ Share a pic of an item that has the same amount of protein you can have
- ☐ Share your daily diet record –completed
- ☐ Share a low-protein meme
- ☐ Share your favorite low protein recipe! Bonus if you cook it and share a pic
- ☐ Dining out, low protein style. Where do you like to eat?
- ☐ Share a pic of what your grocery store haul looks like
- ☐ Real cost of HCU: Grocery Cost Comparison #Medical Nutrition Equity Act, or share some patients with HCU require injectable B12. B12 on average is $300-400 a month and most insurance companies don't cover it!
- ☐ Share a picture or video capturing all the medication you take (this includes formula for those who need it).
- ☐ Share a picture of your first pair of glasses, or a device that helps you navigate or communicate due to lack of vision
- ☐ Share something you wish people understood about HCU
- ☐ #HaikuforHCU—Write and share a Haiku describing life with HCU
- ☐ Wear jeans for your rare genes #ItsInOurGenes
- ☐ Wear your HCU Shirt and share a pic online—#HopeConnectsUs
- ☐ #GoBlueforHCU
- ☐ #HCUAwareness post in a public place
- ☐ Share with a stranger what HCU is and why it's important to you
- ☐ #Create4Cure—Create a work of art that brings awareness for HCU— can be a song, dance, a painting—get creative!
- ☐ #High5forHCU—List 5 ways HCU makes you a stronger, better person
- ☐ All states test for classical HCU, but many are still missed
- ☐ Share a picture of you and a HCU buddy! Or tag a friend who is of great support
- ☐ #FacesofHCU—Share a picture of you saying, I am one of the 1 in 200,000 people with HCU
- ☐ #Hope4HCU—Share 4 things that give you hope and encouragement
- ☐ Share the HCU timeline—if you know other facts, let us know!
- ☐ Cutting Edge of HCU: Share about a therapy that is in the works!

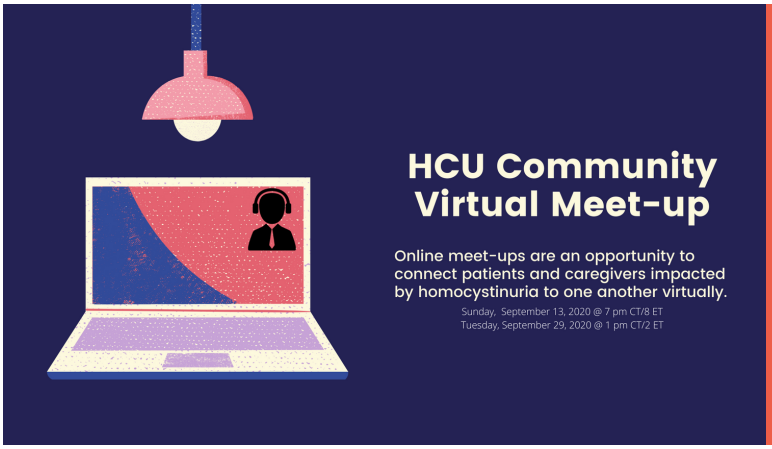
To find additional information and resources, visit:
https://hcunetworkamerica.org/hcu-awareness-month/

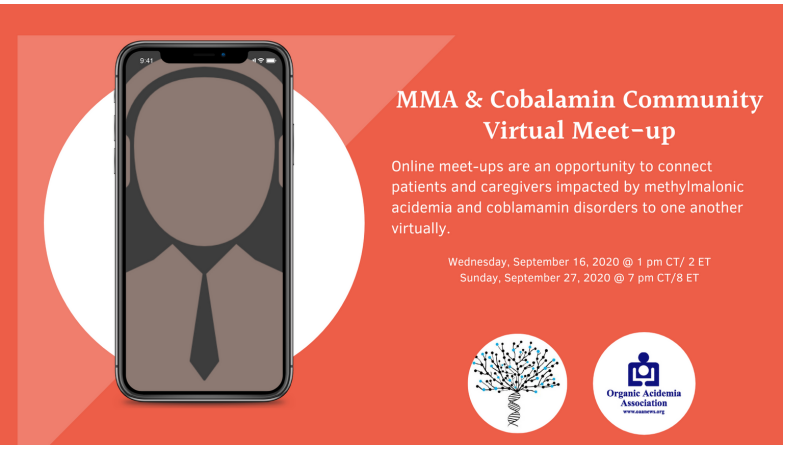
EVENTS

Come check out our Virtual Homocystinuria Meet-ups!

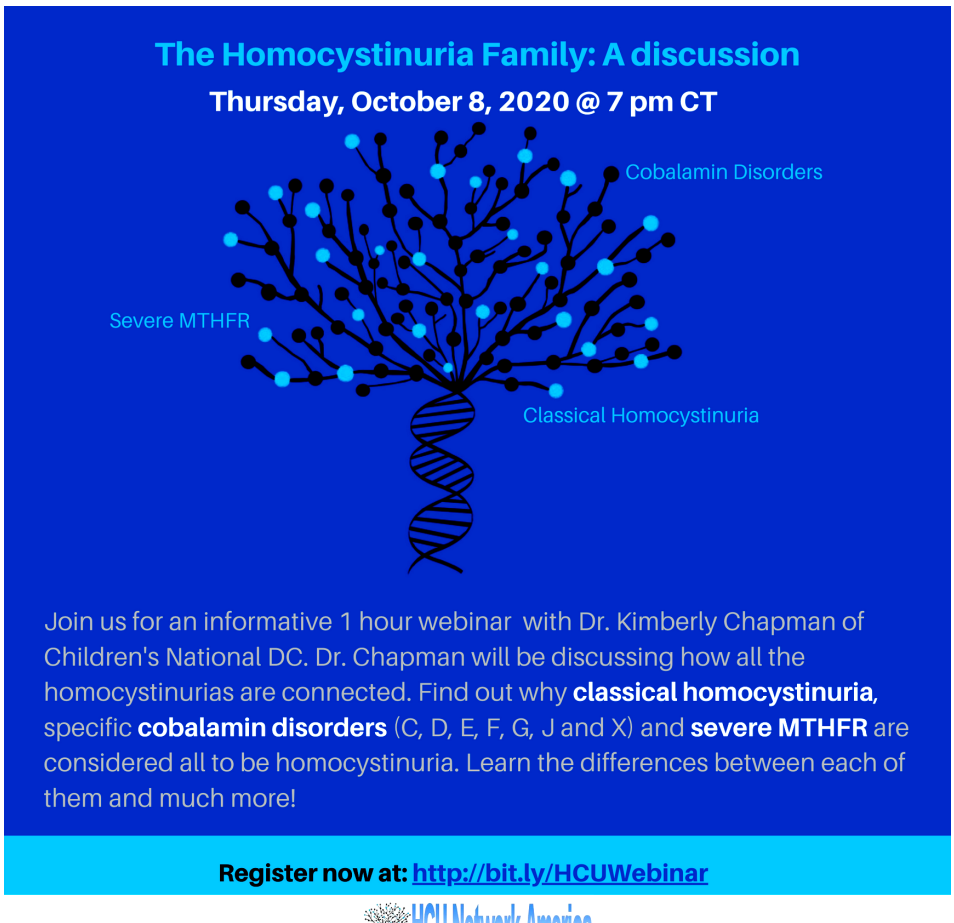
Join our virtual meet-up for a chance to meet, connect, and learn from other patients and caregivers who are facing similar challenges. Whether it's navigating adherence issues, insurance, clinic visit, or life transitions, you are not alone.

Register now at: https://www.eventbrite.com/o/hcu-network-america-30163980100



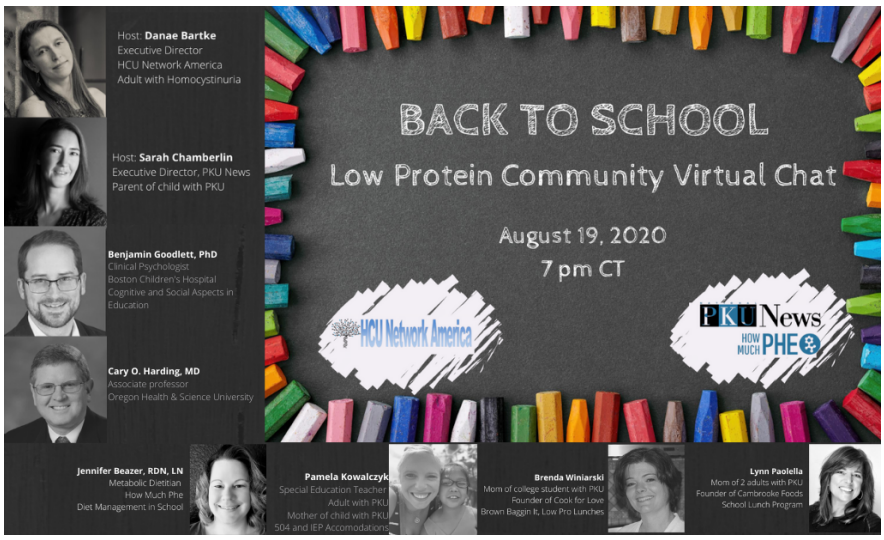


Come take a deeper dive into understanding Homocystinuria - Attend our webinar!



HCU Network America

IN CASE YOU MISSED IT!



Wednesday, August 19, 2020 HCU Network America and PKU News hosted a Back to School Low Protein Community chat that had 6 expert panelist covering topics from Back to School with Metabolic Conditions and Covid, Neurocognitive testing, 504 and IEP plans, The School Lunch Program and Brown Bag Lunch Ideas. This one time, one of a kind of event has been recorded and added to our YouTube Channel. here are also numerous resources in the description section. You can view the recording here: https://youtu.be/Ncp9W0zyqTw

2021 CONFERENCE ANNOUNCEMENT!

HCU Network America and the Organic Acidemia Association are excited to announce the Land of the Free, Home of the Brave, Homocystinuria - Organic Acidemias Family Conference. The combined conference will be held on June 26-27 in Bethesda, MD. The two-day conference will have joint sessions, as well as, individual sessions specific to each patient community.

We hope you are able to join us for this exciting meeting. To find more details please visit: https://hcunetworkamerica.org/2021-conference

CALLING ALL RARE ARTIST

The 2020 Everylife Foundation Rare Artist Contest opened for submissions via Facebook and email on June 16th. Everylife's goal with the Rare Artist Contest is to celebrate the talents of the rare disease community and spread awareness with each piece and the artist behind them. Anyone who is connected to rare is encouraged to enter, they accept entries from ages 4+

Who can enter into the contest?
Anyone connected to the rare disease community can submit artwork, including caregivers, patients, physicians, siblings, and friends. We have three different categories dependent on age; anyone who is ages 4+ can enter. You do not need to be a professional artist or hold any sort of credentials in order to enter into the contest.

Anyone can participate in the voting portion of the contest for the public vote via **Facebook**. We ask that all individuals participating in voting respect the rules. Voters are allowed one vote per art piece every 10 days. You do not have to have a submission in the contest to vote. You do not have to be affiliated with a rare disease in order to vote. The spirit of the contest is about spreading awareness. We encourage you to share your artwork with your rare disease community, to increase your public vote count.

What are the prizes?
Contest awardees are granted cash prizes and an opportunity to showcase their art work at the Rare Artist Reception during **Rare Disease Week on Capitol Hill.** When entering into the contest, we ask that the artwork entered represents the age of the artist. If you are submitting on behalf of someone else, please submit by the age of the artist, not of your own.

Cash prizes according to age groups are as follows;

- Children (4-11) – $100
- Teen (12-18) – $250
- Adult (19+) – $500

Additionally, your artwork will be uploaded to the Rare Artist Website Gallery. Rare Art will also showcase throughout the year at various patient and industry events.

Deadline to apply is 10/01/2020 @ 5 pm EST. Submit your work for the public vote on **Facebook.** For those who do not have a Facebook account, or DO NOT wish to participate in the public vote, please send submission to **lcundiff@everylifefoundation.org**

Family Planning with Homocystinuria

By Katie Sapp, MS, CGC

Once a genetic diagnosis is made in your family, whether by newborn screening or other means, many family members begin to think about the chances for their future children to be diagnosed with the same condition. As a genetic counselor, my job is to help the families I work with understand the condition in the family, who else may have a chance to have a child with this condition, and what options exist to help clarify that chance. Genetic counselors are trained to provide you with information and help you process what a diagnosis in the family means in the context of your life and your values and to help you make the personal decision that is best for you and your family.

If the condition in your family is homocystinuria (which we'll assume it is given where this is being published!), the likelihood for the condition to recur depends on two different factors: how closely related you are to the individual with the diagnosis and the carrier status of your partner. To fully understand how both of these factors impact your likelihood to have a child with homocystinuria (HCU), you first need to understand how this condition is inherited. HCU is inherited in an autosomal recessive manner. Every person has two copies of the CBS gene, one inherited from their mother and one from their father. When both copies of this gene are changed, the body cannot properly breakdown homocysteine and the person is diagnosed with HCU. Because one changed copy of the gene is inherited from each parent, we assume that each parent has a one copy of the gene that is changed and one copy that functions as expected. They are said to be asymptomatic carriers of the condition. For a child to be affected, both parents must be carriers. In this circumstance, each pregnancy the couple has together would have a 25% chance to have the condition.

Once a family member has been diagnosed with homocystinuria, others may choose to learn their carrier status. The closer an individual is to the person diagnosed, the more likely they are to be a carrier. First-degree relatives of the person with HCU (parents, siblings, and children), are more likely to be carriers than those more distantly related. Parents and children of people with this diagnosis are assumed to be carriers of the condition, while an unaffected sibling has a two-thirds or 66% to be a carrier. For anyone related to someone with HCU who wants to know the likelihood that they are a carrier, talk to your doctor or ask to see a genetic counselor and they can help clarify this possibility as well as discuss next steps with you should you want to pursue carrier testing.

Because both parents must be carriers of HCU for a child to be diagnosed with the condition, the carrier status of relatives to the person with the diagnosis is not the only factor to consider when determining the likelihood that a child could also have HCU. The partner's carrier status is equally important when calculating the chance for a couple to have a child with an autosomal recessive condition. Assuming your partner has no family history of HCU, the likelihood that they are a carrier is one in 250 (0.4%). Carrier testing should be pursued for both the partner and the relative of a person impacted by HCU to fully clarify that couple's chance to have a child with the condition.

There are a variety of options available to individuals pursuing carrier testing for HCU. Not all of these options are appropriate for every individual, and not every individual will want to pursue one of these options. The most straightforward approach to testing for the relative of an individual with HCU is called known familial variant analysis. In doing this, you are looking only for the gene changes known to cause HCU in the family. This testing does not look for other changes in the gene, but the likelihood of a relative carrying a different change is quite low, much less than 1%. To complete this testing, your provider would need to have a copy of the gene testing report for your relative with HCU to ensure the correct gene change is assessed. This option is not appropriate for the partner of someone with a family history, as a person from a different family would typically have different gene changes. This testing is often very affordable with some labs offering it for as little as $50.

For family members who do not have access to their relative's genetic testing report or for partners who are interested in carrier testing, gene sequencing is a good option. This testing is essentially spell check of the entire gene. While classic HCU is caused by changes in the CBS gene, there are also other genes which can cause elevated homocysteine levels, so you will want to confirm that the correct gene is being assessed. This testing, while still more expensive than the known familial variant analysis, has come down significantly with many labs offering the testing for $200-$300.

A final option that has become available in recent years in is an expanded carrier panel. These tests were designed to allow for couples to assess what conditions their children may be at risk for, regardless of their family history, by testing many genes (200+) all at once. There are multiple labs offering their own version of this test, but most include the CBS gene for classic HCU. If you're interested in this testing, it would be important to confirm that panel your provider is offering does include the gene for HCU. If it does not, a negative test may be falsely reassuring. Many labs do offer this testing for under $500. It would likely be the most expensive of the options, but it would also yield the most information since it looks beyond just HCU.

If you think you might be interested in pursuing any of these options for carrier testing or simply want more information, talk to your doctor and consider a referral to a genetic counselor. As genetic counselors, we are trained to walk you through all of these options, help determine what information is important to you, and work with your insurance to help ensure you have the lowest possible out of pocket cost. We will also help you interpret the results and talk about next steps for you to consider, as needed. Again, we recognize that carrier testing is not something every person is interested in pursuing. Our job is to help you make the best informed decision for yourself.

If both partners in a couple are identified as carriers of HCU, various options are available to them. Again, in this scenario, the decision a couple makes is deeply personal and should be made by them with their priorities and family in mind. Some couples choose to conceive naturally with the understanding that every pregnancy has a 25% chance to have HCU. From there, some may choose to pursue prenatal diagnosis by either chorionic villi sampling (CVS) or amniocentesis. These tests are invasive to the pregnancy, but they can tell prospective parents with greater than 99% certainty if the baby will have HCU. Other parents will forego prenatal testing and wait until after the baby is born for newborn screening and confirmatory testing to learn the baby's status.

For parents not comfortable with the 25% chance, there are options available through fertility specialists to help lower this risk. Parents can consider using donor sperm from an unrelated

individual or from a relative who is not a carrier. Donor sperm is a more affordable and less invasive option than donor egg or IVF, but it would mean that the father is not biologically the baby's father which can be difficult for some families to consider. Pursuing in vitro fertilization with genetic testing of the embryos by preimplantation genetic diagnosis also minimizes the chances that a baby would have HCU, but it is an invasive and expensive process.

There are many options that can help individuals and couples with an increased chance to have a child with homocystinuria. However, what may seem important or "high-risk" to one person, may not be perceived that way by others despite the risk assessment being the same numbers/chance. The decision to pursue carrier testing, prenatal diagnosis, or other options, is a very personal one. While not everyone is expected to make the same decisions, each person has the right to make an informed decision. A genetic counselor or physician familiar with genetics can help provide you with all the information you need to make the best decision for yourself and your family.

Katie Sapp was also a presenter at our 2019 Homocystinuria conference. You can see a recording of her presentation on this same topic at: https://youtu.be/Lb5BYyGSDkU

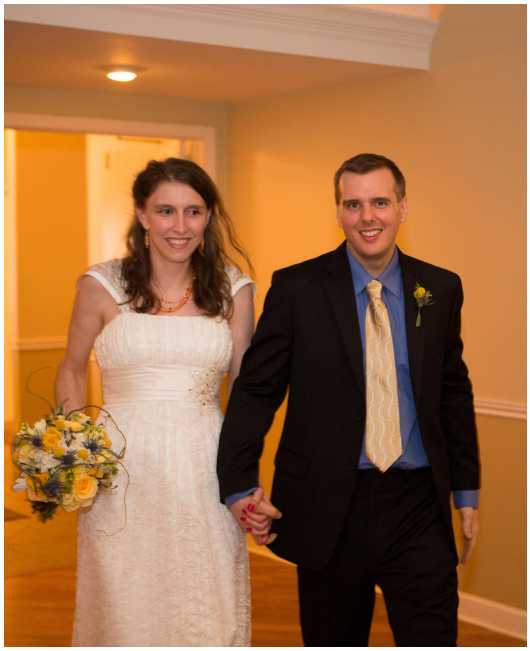
Personal Experience with Pregnancy and HCU

by Danae' Bartke

From as early as I can remember, I knew I always wanted to have my own family. I grew up in a large family; the third of 8 kids. There always seemed to be a baby around, literally!

Growing up, my cousin and I had dreams of moving to Australia and starting our families there. We were going to each have 4 kids and of course 2 sets of twins. So practical, I know. We would practice our fantastic 7 year old mothering skills on our cabbage patch dolls and my younger brothers. We were sure we were going to be fantastic mothers; my brothers might have begged to differ.

In 1995, at the age of 10, I was diagnosed with homocystinuria. We weren't sure exactly what that would mean in the long run for us; we just knew it came with deadly consequences if untreated. As puberty found its way into my life, my geneticists told me I'd likely never be able to have my own kids because of the risks that pregnancy posed to me. Homocystinuria causes an elevated risk for blood clots and so does pregnancy. It was not the news I really wanted to hear because I knew I wanted to have my own family. I eventually accepted that while I may not be able to give birth to my own kids, adoption was still a strong option. By the time I was in my early 20s, the science had started to change and my doctor told me if I wanted to have my own kids I would have to get better control of my homocysteine levels. Since I was still in college, having kids wasn't on my radar and I just brushed it off, knowing if I got my act together I could have kids one day.

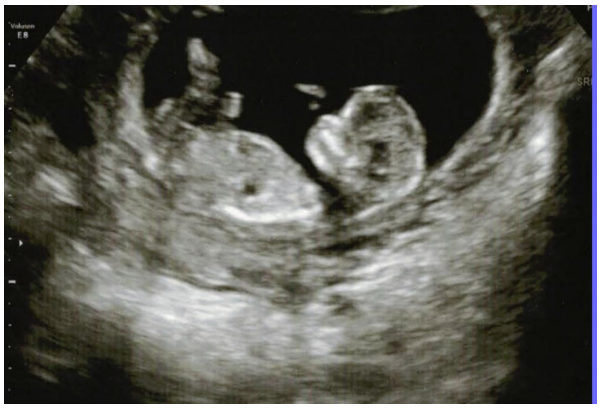


I pursued a bachelor's degree while working full time, not leaving much time to take care of myself. Because of my neglect I ended up having a blood clot. While most people don't view a blood clot as a good thing, I view mine as a life altering experience that put my life on the right path. Not long after my blood clot I started attending low protein cooking classes and metabolic meetings. It really helped me feel part of a community. I met people who had much more strict protein restrictions than myself, and I told myself if they can do it, then I can too! It was the first time I was following the diet and taking my formula the correct way.

In 2011, a couple years after my blood clot, I met the man who would become my husband. In 2014, we got married and shortly after my geneticist requested we see them to get my husband tested to see if he was a carrier. I assured them children were not on our schedule anytime soon, but they insisted – "just in case".

Regardless of the results we knew we would want children of our own, one day but we were a bit relieved when his results came back that he was not a carrier.

In May 2017, well before I was pregnant, I met with maternal fetal medicine specialist and a hematologist to discuss the possibility of pregnancy for a patient with homocystinuria. In August of that year I also started making the necessary steps to switch clinics. The genetics clinic I had been at did not have a team, rather it was just a geneticist. I knew in order to have a successful pregnancy I'd also need a dietitian. I'm very lucky to live in an area with a few options; I ultimately ended up at Ann & Robert H. Lurie Children's Hospital of Chicago. There I had a much more complete team; not only did I have a geneticist, but I had a social worker, advanced nurse practitioner, metabolic dietitian, and genetic counselor.



By the end of October, we found out we were pregnant and my first appointment with the new genetics team was in the middle of November. At my first genetics appointment with the new clinic they reviewed my health history, current diet and formula regime. They increased my formula intake by 15 grams a day as it would not be enough regardless of pregnancy. They took blood samples and got back to me with recommended changes. Because diet and pregnancy are so different, I will not go into detail about the changes they made with vitamins. They did suggest increasing my caloric intake to 2,000 calories from 1,500, which later proved to be too much.

Even with such low total homocysteine numbers the hematologist felt it was important to follow the guidelines for HCU patients and patients who previously had blood clots. At my first appointment at 10 weeks pregnant, they started me on Lovenox injections. About 1 month before my delivery they switched me to Heparin, and then discontinued it 3 days before I was induced. The day following my delivery, they started me back on Lovenox injections, and I continued them for 8 weeks post pregnancy.

Overall my health was very good during the pregnancy. The issues I did have were not believed to be caused by having Homocystinuria. In my second trimester I was told that I had a condition called Placenta Previa. I was very anxious about this because if the condition continued I would need to have a C-section. After my appointment I turned to good old Google and found out the condition was fairly common and 1 in 200 women experienced it and about 75% of the time it corrected itself, so it was very likely I would not need to have a C-section. They continued to monitor the amniotic fluid and position of the placenta until she was born. It did correct itself with time so a C-Section was not needed.

Until our gender reveal ultrasound I was very convinced we were going to have a boy, and my husband was so sure it would be a girl. In February, with our immediate families we announced we were going to have a baby girl. Regardless of gender we had the name picked out, Dana Leah Bartke. Dana after my father, for whom I was named after, and Leah after my grandfather (Lee). In April, between family being in and out of town and my busy conference schedule we planned our baby shower – it was so nice to have everyone together to celebrate the soon to be arrival our of baby girl.

In May, about 1 and a half months before Dana's due date, I was at a genetics appointment and I mentioned to them how swollen and itchy my feet had become. They were so enlarged I could no longer fit in my regular shoes and the only comfortable shoes I could wear were sandals, mostly flip flops. The swelling wasn't so bad, but the itchiness felt unbearable! Nothing I could do or take helped. I tried soaking them in hot water (later found out that made it worse) then iced them – that helped for a little bit. I tried Benadryl cream, that would help, but ultimately would end up itching again just minutes later. My genetics team found this very concerning and ran some test. While I was at my other doctor appointment later that day I got a call telling me I needed to go to the hospital for further blood work and to be monitored overnight, that they suspected a condition called Intrahepatic cholestasis of pregnancy, or just commonly known as cholestasis of pregnancy. The next day I received confirmation that I did indeed have this condition. This meant being induced at 37 weeks, because anything past that was dangerous to the health of the baby. They sent me home with medication to take daily and then had me followed by Maternal Fetal Medicine more frequently (twice a week instead of once).

At exactly 37 weeks to the day, they had me check into the hospital at 6 am and started the process of being induced. The first day was pretty non-eventful, by 10 pm I hadn't made it past 4 cm so I went to bed hooked up to a plethora of monitors and I Vs. The next day continued to also be non-eventful. At about 9 pm they came in and broke my water, which quickly sped up the process. With each passing minute the contractions picked up getting me closer and closer to having her. At 10:38 pm Dana finally was born!

This next part may be a bit scary for some, but I feel it's an important part to share. Minutes after Dana was born, I started to have additional issues. They could not get the placenta to detach, and along with the difficulty I started hemorrhaging; my uterus was not contracting properly. They immediately started me on medication to stop the hemorrhaging and started a blood transfusion. I was in a pretty weakened state of things, so the first few hours I spent under a heating blanket and Dana was in the nursery – not how any mother anticipates the first moments/hours of their child's life. Most importantly though, Dana was healthy – even at 37 weeks she weighed just under 8 lbs. I later learned that roughly 18-25 percent of births involve a postpartum hemorrhage – something they do not talk about at all in any birthing or pregnancy class.

Four days after Dana was delivered both of us were cleared to go home. We finally got to be a family of three in our own home. Life has definitely been an adjustment. My homocysteine levels continue to stay down around 15-17 and immediately after Dana was born they had me go back to 20 grams of protein a day. I had to reestablish new routines for my formula and eating that worked with having a baby – I didn't anticipate that being so difficult, but it's doable.

Today, Dana is a now a very typical 2 year old. She is talking, running, and into everything. She loves baby

dolls, bunny rabbit stuffed animals and loves to sing. Some people have asked despite the cholestasis and my delivery experience would I do it again, yes – and eventually we do plan to have a second.

My advice if you have Homocystinuria and want to have children is this:

1. Get your levels down into safe range – whatever your doctor deems that is. Some might say 50, some might say 25 – it's doable!
2. Get your partner tested to see if they are a carrier. As Katie shared in the previous article there are a lot of options – in our case we were going to have our own children either way.
3. Make sure you have a team you feel can help guide you through your pregnancy – not just a geneticist, but also a dietitian and a MFM that is familiar with Homocystinuria.
4. Enjoy the time you are pregnant! Things may come up regardless if you have Homocystinuria or not.

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 #2020NBS
- Add the #2020NBS frame to your profile picture
  a. Update your profile picture
  b. Click add frame
  c. Search for #2020NBS
  d. Reposition your profile picture accordingly
  e. Determine how long the frame will be displayed

Raise Awareness on Twitter

- See example Tweets: https://www.babysfirsttest.org/newborn-screening/2020-newborn-screening-awareness-month
- Attend the twitter chat!

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?
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.

On the following page you will find the letter portion. We ask you to give to your clinic, followed by the survey form:

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_____

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
Or complete the survey online: https://hcunetworkamerica.org/survey-on-classical-homocystinuria-patients-missed-by-newborn-screening/

Why Do Families Need Navigate Newborn Screening?

Newborn screening (NBS) is a health screen that checks for serious conditions at birth. NBS is a life-saving service, available to the nearly 4 million babies born in the United States each year. To better understand family preferences for NBS education, Expecting Health surveyed **819 participants** made up of parents, expecting parents, individuals with NBS conditions, or family members of individuals with NBS conditions.

LIMITED NEWBORN SCREENING AWARENESS

2 out of 3 participants are aware of NBS.

1 out of 3 participants aware of NBS can correctly identify a definition of NBS.

DISPARITIES IN NEWBORN SCREENING EDUCATION

Participants living in HRSA-defined medically underserved areas (MUA) may experience disparities in NBS education compared to those living in other areas.

55% of participants in an MUA were previously aware of NBS compared to **67%** of those living in other areas.

50% of participants in an MUA learned about NBS before birth - the optimal time - compared to **61%** of those in other areas.

FAMILY LEARNING PREFERENCES

An **online module** was the preferred format compared to other educational formats.

Family stories were considered very helpful to learn about NBS.

Participants use **social media** to connect with others about NBS and other health topics.

Informed by this data, Expecting Health developed ***Navigate Newborn Screening***, a free online learning module that helps families just learning about newborn screening and provides opportunities to become leaders in the newborn screening system.

Sign up today: https://expectinghealth.myabsorb.com?KeyName=NavigateNBS_HCUNA

Have questions? Contact **Annie Evans at aevans@geneticalliance.org**

Navigate Newborn Screening
An Expecting Health Program

This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling $400,000 with 0 percent financed with non-governmental sources. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS, or the U.S.Government.

Navigate Newborn Screening

An Expecting Health Program

WHAT IS NAVIGATE NEWBORN SCREENING?

Navigate Newborn Screening is a free, learning opportunity that gives families information on one of the most common tests newborns get - newborn screening. The module can help families just learning about screening as well as those looking to be leaders in this system.

In this module, you will learn about:

- The newborn screening process
- Newborn screening results
- Types of conditions detected
- Questions to ask your healthcare provider
- How to tell your newborn screening story
- Additional newborn screening resources

WHY LEARN ABOUT NEWBORN SCREENING?

- Newborn screening is **a state-run public health service** that ensures all babies are screened for certain conditions that can cause serious health problems.
- Newborn screening usually happens when your baby is between **24 and 48 hours.**
- In the U.S, all states require newborn screening, but **not every state screens for the same conditions.**
- **Only 1 in 3 people** can correctly identify the definition of newborn screening.

BENEFITS OF PARTICIPATING

Learn about the most common screening test

Gain leadership and advocacy skills

Options to attend national conferences or meetings

Sign up today https://expectinghealth.myabsorb.com?KeyName=NavigateNBS_HCUNA

Interested but have more questions? Contact **Annie Evans at aevans@geneticalliance.org**

Expecting Health

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Live better, together!

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/

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