HCU Herald

Presented by HCU Network America

Connecting for a Cure.

There have been a lot of things happening for the HCU community & for HCUNA.

We strive to keep you informed and connected.

Inside this issue:

Heroes of HCU

Maeve From Connecticut

Maeve was born in May, 2012. As part of the check-in process and welcome procedure, she had her heel pricked and blood drawn for testing. We didn't know at the time that the state of Connecticut tests newborns for several different genetic and metabolic conditions, so we were surprised when our doctor told us to come back in to meet with a geneticist, that she had tested positive for HCU. We would come to be very grateful for these tests.

At first we were confused, and more than a little scared. There is very little information out there about HCU. We didn't really know what to do or what to expect. With the guidance of our pediatrician, geneticist, and dietician, we soon began to understand what our daughter could look forward to, and what our daily routine would be like. We also found a tremendous resource in the stories of others with the condition.

Hearing first hand reports from other parents and people helped us navigate how to adjust our lives to accommodate the condition. In short, the devastating effects of homocystinuria can be avoided through long-term dietary management. By eating foods that are naturally low in protein, as well as low in methionine, the body can maintain normal levels of the metabolized proteins. If those proteins were to build up over time, the results would be toxic and possibly fatal. The first step for us, though, was to determine whether Maeve would respond to a vitamin B6 supplement.

After we determined that Maeve was B6 non-responsive, we began weekly blood draws to monitor her homocysteine levels. Her diet became a trio of substances: her normal baby formula; ProPhree, which is a powder formula containing no protein - only minerals and vitamins; and Hominex, a medical food whose protein is broken down into a state that an HCU patient can process. The approach was that she would get 80% of her daily protein needs from medical food, and the remaining 20% from "intact sources", or normal food. We planned that she would be able to have about 5 grams of protein per day, with anywhere between 150-200mg of methionine.

At the beginning, it was fairly easy. We carefully measured out the powder to mix with her formula and water. We were cautious not to spill any of her bottles, as they had precise amounts for her nutritional needs. This became a delicate proposition when Maeve wanted to feed herself. With a lot of patience, and a very attentive eye, we were able to let her hold the bottle and feed herself without too much food being lost.

Soon after that, we had to figure out solid foods. We knew that we'd be sticking pretty close to fruits and vegetables, and the baby food purees were great. We knew exactly how many calories each jar contained, and how many grams of protein she got. It was very easy to track. We were worried that she'd get stuck on the texture, and not be willing to try different foods.

As luck would have it, once her front teeth came in, she discovered the joys of crunchy foods. We found that the baby food designed to encourage self-feeding works out great. There are puffs and cookies and crunchies that are all zero-protein, and they add to the calorie count we're trying to meet. She also loves to eat melon, grapes, lettuce, cucumbers, and even some green peppers. We've found some all-veggie snack chips that are made with powdered tomatoes. She just loves to crunch on those! Along with the food, we give Maeve a medicine called Betaine to lower homocysteine by metabolizing it back to methionine. The levels are being adjusted along the way, but she is currently receiving a daily dose of 1950mg. They come in capsule form, and we empty the contents into her formula throughout the day.

One of the things we read after Maeve was born was the importance of keeping a log. This lets the sleep-deprived parent keep track of the newborn's activity. We've kept this habit up, and it's made it easier to keep track of her progress as she grows. We've taken the logged information about how many calories she's taken in each day and put it into a spreadsheet. This lets us go back and chart her progress over time. It's made it really easy to see when she's been sick with a cold, for example, and hasn't been eating normally. We also use it to track against her blood levels, and working with the nutritionist we're able to adjust her diet to compensate for teething, colds, and other trends that may not be apparent from just the blood test results.

Now Maeve is almost two, and everything is going great. Maeve is growing like a normal kid. She's very tall for her age, and she seems to have a never-ending supply of energy. She is happy, and loves to read and play. Her blood levels are well within the safe range, and though they fluctuate a little bit, we're becoming quite comfortable following the guidelines of her diet. We initially thought the HCU diagnosis would be difficult and problematic, but it hasn't been tough as we expected. Aside from the normal problems that all parents have convincing their kids to eat, it hasn't been that big of a deal.

HCU and You: *Ask Methia*

DEAR Methia,

How do I get my toddler to drink there formula again?

We are parents of a toddler with HCU, and we are in the process of transitioning him to a toddler-appropriate metabolic formula. This transition has been tough. He would rather drink water and juice, and has actually begun to throw his sippy cup of formula across the room when given to him. When he was a baby, he took his formula in minutes without any difficulty. Why is this happening, and what can I do to make sure he gets what he needs?

Love,

A Family in Formula Frenzy

Dear Family,

You are not alone! Transitioning from a bottle to a sippy cup alone can be tough, and when complicating that transition further with a formula that doesn't always taste great, it can be a recipe for long mealtimes and headaches. Here are some tips that might help.

- **Try some flavor enhancers.** We know that these products don't taste like a milkshake, but we can try our best to make them more palatable! Try complimenting flavored formulas with products that might mask the taste, and transform it into a better tasting product. For example, a vanilla flavored formula might be complimented well by a splash of orange flavoring to make a "creamsicle," and the aftertaste of a berry formula might be altered for the better with a dash of fruit punch. Mio, a newer product on the market, can flavor formulas with just a few drops of naturally sweetened (and zero calorie) liquid.

- **Make drinking formula fun.** Instead of formula being something that becomes a fight between you and your child, turn it into a game. Imagine – suddenly, the HCU formula has transformed into a "Power Drink" that will give them super-human energy! Or, drinking 100% of daily formula will let your child earn a sticker on a calendar, and a whole week's worth of stickers will earn them a prize. Creativity is key.

- **If the formula isn't working out, switch!** Ask your metabolic dietitian to have formula samples sent to your home. They are knowledgeable about new products on the market and can make recommendations about what might be best for your child to try. Sometimes, when transitioning formula, mixing "the old" and "the new" formula together initially and slowly switching from one to the other works best to avoid the shock of something completely different. Reach out to your clinic and ask for suggestions if nothing seems to be working to improve compliance on the current formula.

Remember, your clinic is always available to you to help you troubleshoot these situations, and can also connect you with other families who have most definitely gone through the same struggles!

Sincerely,
Methia

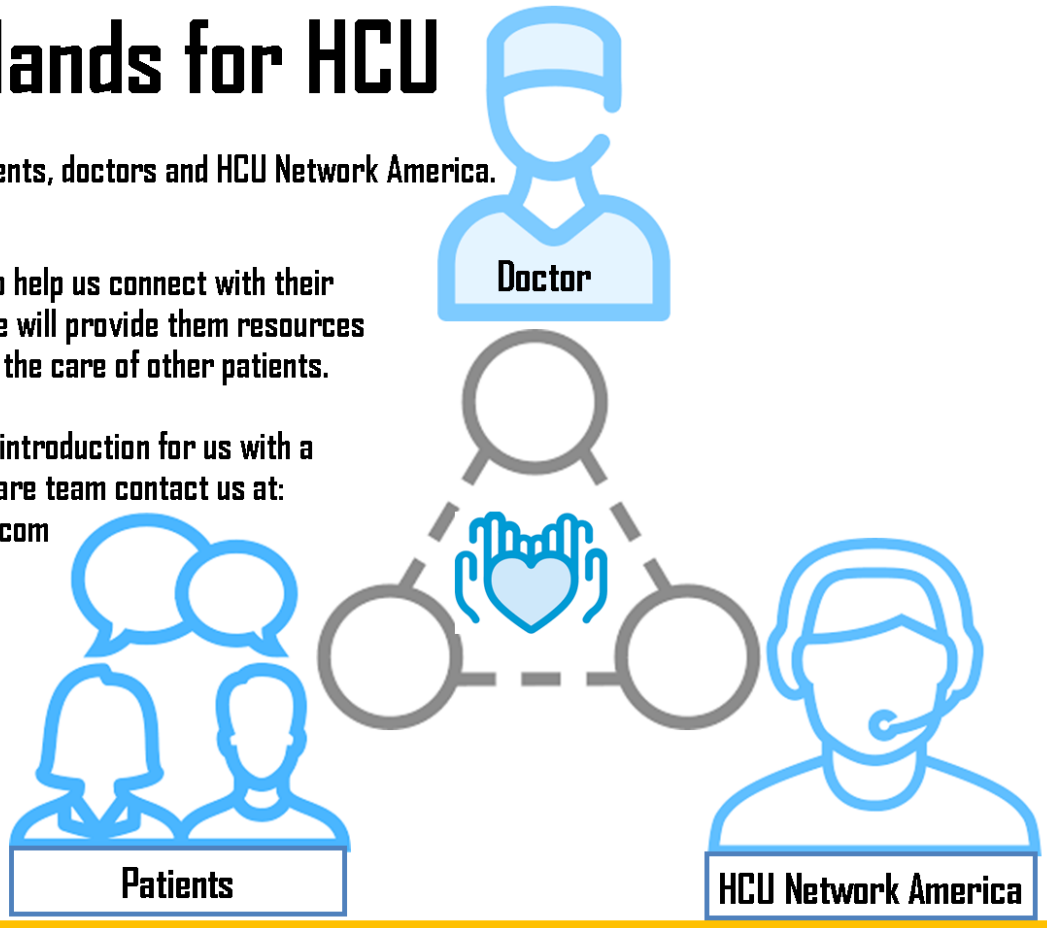
Ways to Get Involved

Helping Hands for HCU

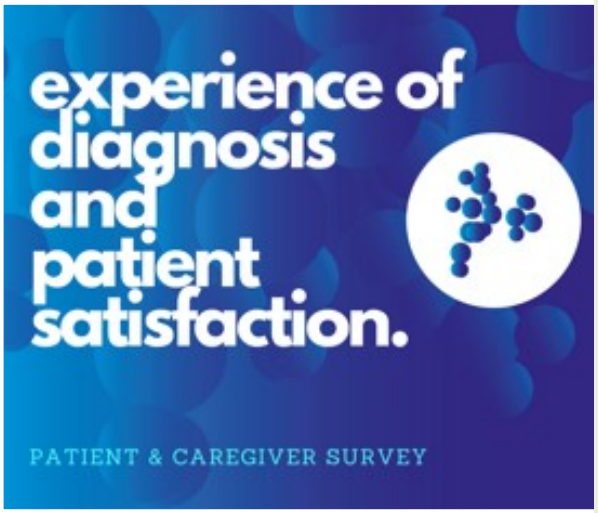
A collaborative effort of patients, doctors and HCU Network America.

We are asking for patients to help us connect with their metabolic team. In return, we will provide them resources to help better your care and the care of other patients.

If you are willing to make an introduction for us with a member of your metabolic care team contact us at: HCUNetworkAmerica@gmail.com



HCU Network Australia Patient & Caregiver Survey



There is currently limited evidence showing the considerable length of time taken in some cases to reach a diagnosis of homocystinuria and the level of patient satisfaction regarding access to information, treatment options and medical care. This survey is important to highlight the current situation and has been prepared in consultation with a Key Opinion Leader (KOL) in Europe together with input from HCU Network America, to ensure the information gathered can be used globally to help support improved care for all HCU patients.

We ask no matter what your experience, good or bad, you complete the survey.

To complete the survey click here.

Natural History: Indiana University

Orphan Technologies is pleased to announce that it is expanding its Natural History Study sites!

Orphan Technologies has just signed an agreement with Indiana University School of Medicine in Indianapolis, Indiana to include them in the Natural History Study. They are the first site in the Midwest to join the study. More sites are in the works!

The study does not involve any investigational medications, but will provide information to researchers who are currently developing a medication to treat the disease.

The study has 5 main parts, all of which are provided free of charge to participants: bone exam (DXA scan), blood draws for important health tests, comprehensive eye exam, cognitive testing, and physical exams.

Here is a list of current sites:

Emory University, School of Medicine Department of Human Genetics
Decatur, Georgia, United States 30033

Indiana University School of Medicine
Indianapolis, Indiana, United States 46202

Boston Children's University
Boston, Massachusetts, United States 02115

The Children's Hospital of Philadelphia
Philadelphia, Pennsylvania, United States 19104

To learn more about the study, please click the link below:
https://clinicaltrials.gov/ct2/show/NCT02998710

Or contact:
Anne Kuan
(781) 966-3832 or anne.kuan@neovii.com

Contact Register

Did you know that just under one year ago we launched our 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/

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Please visit and encourage others, too!

Coming Soon: HC&U Podcast

HC&U

A PODCAST ABOUT HOMOCYSTINURIA

Current research

Interviews

Low protein Recipes

Day to day life

And many other topics!

COMING AUGUST!

What is HC&U?

HC&U is a podcast about Homocystinuria, sponsored by HCU Network America and hosted by Ben and Lindsey Massengale. Ben was diagnosed with HCU at birth, and Lindsey had no idea what HCU was. Then Lindsey met Ben, they got married, and Lindsey had to learn how to cook vegetables in a bajillion different ways. This podcast will include interviews with different professionals in the field, a low protein recipe each episode, and even personal stories from Ben's experience with HCU. We are open to suggestions on topics you would like to hear, so please email us any ideas you have. Watch our social media sites for the podcast release date!

HC&U

Twitter: @hcupodcast

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HC&U: A Homocystinuria Podcast

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We'd like to thank the following content contributors:

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