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.

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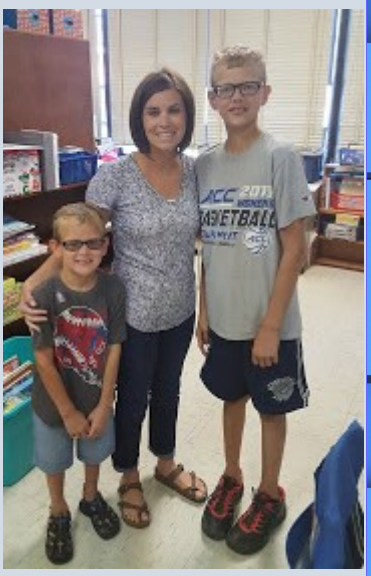
HCU Heroes:
Zak and Gavin from North Carolina

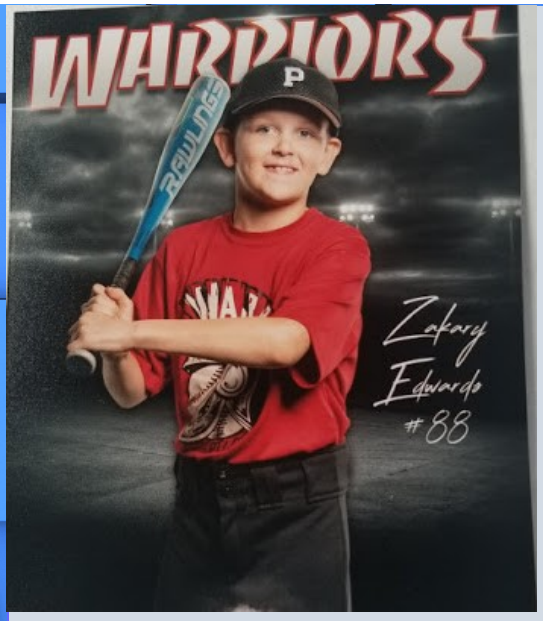
My name is Amanda and I have four wonderful boys and two of them were diagnosed with HCU in July of 2016. We found out when Gavin went for his annual eye exam the end of 2015 that his lenses had dislocated and just a month after that we took Zakary for his annual eye exam and found out that his lenses were also dislocated. They referred us over to a geneticist at Brenner Children's Hospital and that is when we found out that they had HCU. The geneticist at Brenner's wasn't experienced in HCU so she sent us to UNC Children's Hospital to see Dr. Lori Smith. When she did blood work and got their levels back they were both over 300.

We have battled constantly with their levels due to having a hard time with adjusting to the food change and the medicines. Since Gavin was 10 and Zakary was 5 when they were diagnosed they didn't take well to the formula and diet change. In March of 2017, Zakary was sent to Nationwide Children's Hospital in Columbus Ohio to have his lenses removed and artisan lenses put in place, and in August 2017 Gavin had the exact same surgery. In total we had to make over 20 trips in a year.



Then in July 2018 Zakary had an accident at home playing with his little brother and got one of his lenses knocked loose so we had to make an emergency trip to Ohio to have another surgery to repair it. After these surgeries the boys made 100% turnaround with their vision. Before their surgeries they both couldn't see the 20/400 letter with glasses and after their surgeries they both could see 20/30 and 20/40 without glasses! We were so excited of the progress that both the boys were making. Their levels were slowly coming down and everything was looking up for them.

Then sadly on Thursday October 19th, 2018 Zakary had just finished playing his baseball game and about 2 miles from the ball field on the way home, Zak told me he was going to be sick so I pulled off the side of the road and he got out. He never got sick and got back in the car and started jumping up and down and had tears rolling down his face and told me to call the doctor. I got him back out of the car and he was breathing really fast and still jumping and crying. I looked over at him and his face had turned blue. My husband Brian pulled up right after that and started CPR. A nice man had stopped to help me and told us to take him to the fire department which was about a mile from us. Brian loaded him in his truck and we took him to the fire department. He had already went limp again so he started CPR on him again. I finally got the firemen out there to help us and shortly after that the EMS arrived and took over. While in the ambulance, the paramedics lost him for 15 minutes while on the way to the hospital. Once they made it to the hospital they had finally got him stabilized and things were looking ok, then he started having continuous seizures that



they couldn't get under control. The first few days everything was looking good. His brain wasn't swelling and his eyes were still reacting. He was out breathing the ventilator. Then Saturday came around and he still wasn't waking up and nothing was really changing, and the doctor called us into a small room and told us that things weren't looking good and that we most likely would be going home without him. We tried to stay positive because we knew the only person that could tell us that he wouldn't wake up was God. Saturday night came around and they did another CT scan and his brain had swelled so bad that you couldn't even make it out. It was just solid white. Monday his eyes stopped reacting and that is when we knew things weren't looking good. On Wednesday October 24th 2018 our precious Zakie left us to go play baseball with the angels in the outfield.

It has been almost 3 months now that he gained his angel wings, and it still feels like a bad nightmare that I'm going to wake up from and see my little boy running at me with arms opened wide and that big smile saying mommy I love you and giving me one of his big bear hugs. The hardest thing for me is that I will never get to cheer him on at his baseball games (which were his life), I'll never get to see him graduate high school, I'll never get to see him get married or have children. Our community has been a wonderful support and it just shows how many lives that our sweet angel touched. We have had so many people to tell us that they had only met him once but that he had made such an impact on them. He had a pure heart and loved everyone no matter your flaws. He never saw anything wrong with anyone. When we were at church he would always be the first person behind you if you went to the altar to pray and he always put everyone before himself. His 3 brothers have taken it really hard especially his little brother, Hunter. He was his best friend they did everything together. Hunter talks about Zakie everyday, no matter what.

Needless to say our journey with HCU has been very traumatic and I pray that I won't have to ever deal with something like this again, but knowing that Gavin also has HCU scares me to death because we never know if something like this is going to happen to him also. The only thing we can to is to work harder at getting Gavin to follow his diet and take his formula like his is supposed to. I do believe that if we would have found this on their newborn screening it would have been a lot better for both of the boys in many ways. First, they would have taken their formula without having as many problems because they would be used to it and second, the diet wouldn't have been a big issue because they wouldn't know any different.

This has been a long journey in just a little over 2 years, and I would have never guessed that we would have had so much to go through in such a short time. I just pray that they will be able to find a way to catch HCU better on newborn screenings so that it gives the children a better chance of life. Both Gavin and Zak had several side effects from not finding out until they were older. They both had a severe learning disability and they both had to have the eye surgery. Zakary had a mild case of scoliosis and they both had several behavior problems from it.

This letter has been very hard to write but I know by telling our story that we may be able to help another family from having to go through what we have been through. To read more about the Edward's Family story, you can click on the following link: thomasville-parents-mourn-loss-of-8-year-old-son-who-had-rare-genetic-disorder

New News: Meet Angela Pipitone, Medical Advisor

Angela Pipitone is a metabolic dietitian at The Johns Hopkins Hospital in Baltimore, MD, United States. Angela specializes in medical nutrition therapy for inborn errors of metabolism, including homocystinuria and other methylation defects. She holds a Bachelor of Science degree from the University of Delaware with a major in Dietetics, minoring in Psychology. She went on to complete her dietetic internship with a clinical focus at Johns Hopkins Bayview Medical Center in Baltimore, MD. Licensed and registered to practice as a dietitian in the state of Maryland, she began her career in 2009 at Suburban Hospital in Bethesda, MD, as a clinical dietitian in the adult intensive care unit. The realization that she wanted to specialize and work with both the pediatric and adult population led her back to Baltimore in 2013, when her passion for working with and treating metabolic patients began.

Angela is a constant on the Hopkins metabolic service, providing medical nutrition therapy for inpatients in metabolic crisis and continuity of care for these same patients in the outpatient clinic. She firmly recognizes the importance of patient-specific diet treatment and education, noting that all metabolic patients within the same diagnosis are uniquely different. She enjoys working with patients and families to problem-solve issues that arise surrounding diets and formulas, and eagerly attempts to "make food fun." Angela also enjoys writing, both for research purposes and for fun, and loves unleashing her creativity under her alias, "Methia," for HCU Network America.

HCU and You: *Ask Methia*

DEAR Methia,

How Can I Make School Lunches More Exciting?

Dear *Methia*: My daughter with HCU is finishing the 6th grade this year. I just returned from her last parent teacher conference and learned that she has been eating her lunch in the nurses' office every day. When I approached her about this, she told me that all of her friends buy their lunches, and were always commenting on her "boring packed lunch," which is the same every day. To avoid their comments and questions, she decided to sit alone in the nurses' office because that is where she goes to take her formula. This hurts me, because I remember lunch hour being one of the best parts of my day as a kid. What can I do to help her during the next school year?

Sincerely,
Lonely Luncher

Dear *Lonely Luncher:*

The struggle to avoid boredom on a low protein diet is SO real! Following a diet that is significantly restricted doesn't lend to too many food options, and often requires that people with HCU eat the same foods on repeat. We call this "food boredom." Not only does food boredom serve as a barrier to compliance, it makes eating seem like a chore rather than an enjoyable experience. Fortunately, there are ways for you to help (and teach!) your daughter when it comes to making food fun and more creative.

Involve your child in weekly meal planning. Trips to the grocery store make meal planning much more fun, particularly when your child has a say in what is purchased! This activity also allows you both to explore new food items that are available. Practice reading food labels and think about ways that these new foods can be incorporated into meal plans. After these trips, you can sit down together and put together a few "sample meals," amounting to about the same amount of protein, so that your child has a choice as to what is in her packed lunch.

Ask for your metabolic clinic's assistance with the School Lunch Program. The Americans with Disabilities Act Amendments Act of 2008 made important changes to the meaning and interpretation of the term "disability." The definition now includes anything that substantially limits a major life activity. Certainly, having homocystinuria affects eating – a major life activity! School food authorities are required by law to provide modified meals for people requiring low protein diets. Your geneticist and dietitian are able to write letters of medical necessity for these mealtime modifications, and can even write for the use of low protein medical foods. Your dietitian can then work with the school to develop a cycle menu for your child, with your input, that is both satisfying and compliant!

Remember, your clinic is always available to you to help you brainstorm solutions to your concerns. They are not only your best resource, but a gateway to becoming connected to others who can help!

Sincerely,
Methia

Back to School: Webinar Series

Save the Date

Homocystinuria Community
Tackling the School Meal Dilemma

Join Cambrooke's Founder, Lynn Paolella, and HCU Network America Executive Director Danae' Bartke, for a 1 hour webinar:

May 8, 2019

12:30-1:30 pm EST

WEBINAR OBJECTIVES

- Your child's 'Civil Right' in the National School Lunch Program
- Make the process easy to understand
- Provide information, tools, and resources to help your child "Join the Lunch Line"
- Brown bagging it creatively

DISCUSSION TOPICS

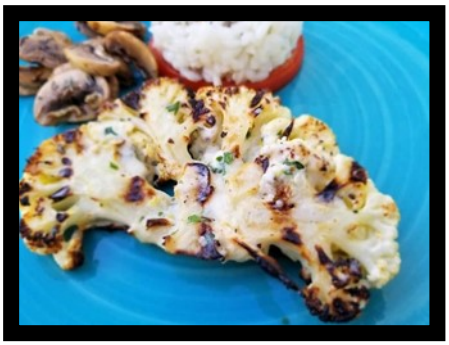
- What is "Dietary Accommodation"
- How to "Communicate with your School"
- Personalize Program to YOUR child

Join Us!

AJINOMOTO. CAMBROOKE THERAPEUTICS™ HCU Network America

CLICK HERE TO REGISTER

Recipes from the Kitchen



Grilled Cauliflower with Garlic Herb Compound Butter

Author: Amber Gibson

Makes 2 Cauliflower Steaks
Serving: 1 Filet
Protein: 2 g per filet

Ingredients:

- 180 g Cauliflower Steaks, about 2 filets 1 inch thick each
- 1 TBSP Oil, Olive
- 1 TBSP Oil, Canola
- 2 TBSP Cambrooke Seasoned Breadcrumbs
- 1 TBSP Garlic Herb Butter (See recipe below)

Instructions:

Preheat your grill to medium high heat. Combine the olive oil and canola oil in a small bowl. Brush oil over the cauliflower steaks and season with salt and pepper. Gently coat with Cambrooke's seasoned bread crumbs. Place the cauliflower on the grill and cover. Cook for 10 minutes. Flip steaks and grill another 5 minutes. Brush the garlic herb butter over the steaks and continue to cook for 5 more minutes. Remove and serve immediately.

Garlic Herb Compound Butter

Ingredients:

- 2 TBSP Cambrooke Garlic n Herb Cream Cheese, softened
- 1 TBSP Sour Cream
- 4 TBSP Butter, regular or unsalted, softened
- 1 tsp Lemon Peel (zest), fresh
- 1 tsp Lemon Juice
- 1 tsp Parsley, fresh, chopped
- 1/8 tsp Salt, Table
- 1/8 tsp Pepper, black

Instructions

Place the softened garlic herb cream cheese, softened butter, cream cheese, lemon peel, lemon juice, parsley, 1/8 teaspoon salt, 1/8 teaspoon pepper. Use a spoon to mash and blend all the ingredients together. Refrigerate until ready to use.

Makes 115g total
Serving: 1 TBSP or 15g= 0.2 g protein

Recipes from the Kitchen

Grilled Sweet Potatoes with Herb Butter

Author: Amber Gibson

Makes 5 servings
Serving: 2 planks
Protein: 1.7 g protein per serving
Calories: 175 per serving

Ingredients:

- 500 g Sweet Potato, about 1 large
- 2 TBSP Olive Oil
- 1/2 tsp Salt, Table
- 1/4 tsp Pepper, black

Instructions:

1. Preheat grill to medium heat.

2. Peel sweet potato and cut crosswise. Cut into 1/2 inch thick planks. Place the planks in a medium bowl and toss with the olive oil, salt, and pepper.

3. Place the planks on the preheated grill and close the lid. Cook for 5 minutes, then flip to grill the other side. Cook for another 5 minutes. Use a basting brush to spread the softened herb butter over the grilled sweet potatoes. Continue to cook until sweet potatoes are fork tender. Remove from grill, brush with some more butter and serve immediately.

Herb Butter

Ingredients:

- 4 TBSP Butter, unsalted, Softened
- 1/4 tsp Garlic Powder
- 1 tsp Parsley, fresh, chopped
- 1/2 tsp Basil, raw, chopped

1/2 tsp Chicken-Flavored Consommé & Seasoning, dry

Instructions

In a small bowl add the softened butter. Mash the butter with a fork until smooth and creamy. Next add the garlic powder, parsley, basil, and consommé until well blended. Place into a sealed container and refrigerate until ready to use or you can use it right away.

Recipes from the Kitchen

Grilled Watermelon Filet

Author: Amber Gibson

Makes 2 servings
Serving: 1 Filet
Protein: 0.8 g per filet

Ingredients:

- 200 g Watermelon Filets, 1 1/2 inch by 4 inch rectangle cut and about 1inch high
- 2 TBSP Steak Sauce
- 1/2 tsp Salt, Table
- 1/4 tsp Pepper, black

Instructions:

1. Preheat grill over medium flame. Lightly spray the watermelon filets with nonstick cooking spray and place on the grill. Allow to cook, without moving the filets, until you see nice grill marks. It should take about 5 minutes. Flip the filets with tongs.

2. While cooking the other side, baste the filets with the steak sauce and season with salt and pepper. Once you see nice grill marks on the other side, flip again and quickly baste this side with the steak sauce and season with salt and pepper. Remove from grill and serve immediately with low protein rice, mashed potatoes, roasted vegetables, etc.....

Go the Extra Mile for HCU: Virtual Event!

Go The Extra Mile For HCU

This May, by joining HCU Network America for their first **Virtual** Race

500 Miles (combined) for HCU - in and around the Indianapolis Motor Speedway

We're working with virtual race platform Racery to bring you the first annual HCU virtual race! While Racery offers lots of fundraising virtual challenges for charities, including a virtual English Channel swim and Everest virtual stair climbing challenge, we decided to build our own virtual race to lead into our Indianapolis conference.

What is a virtual race?

A virtual race is a race that can be walked, run, biked or bladed 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.

How do you know how many miles I completed?

1. We rely on the honor system. You don't have to use a device to prove your miles. If you'd prefer to use an app to track your miles, we recommend Strava. You can join the HCU Network America Club.
2. Please use intentional miles - this means no step counting
3. Please attribute your activity to the appropriate day -- don't combine workouts from multiple days.
4. Please only backdate logs for a race.
5. Activities submitted after the race ends will not count toward race leader boards.

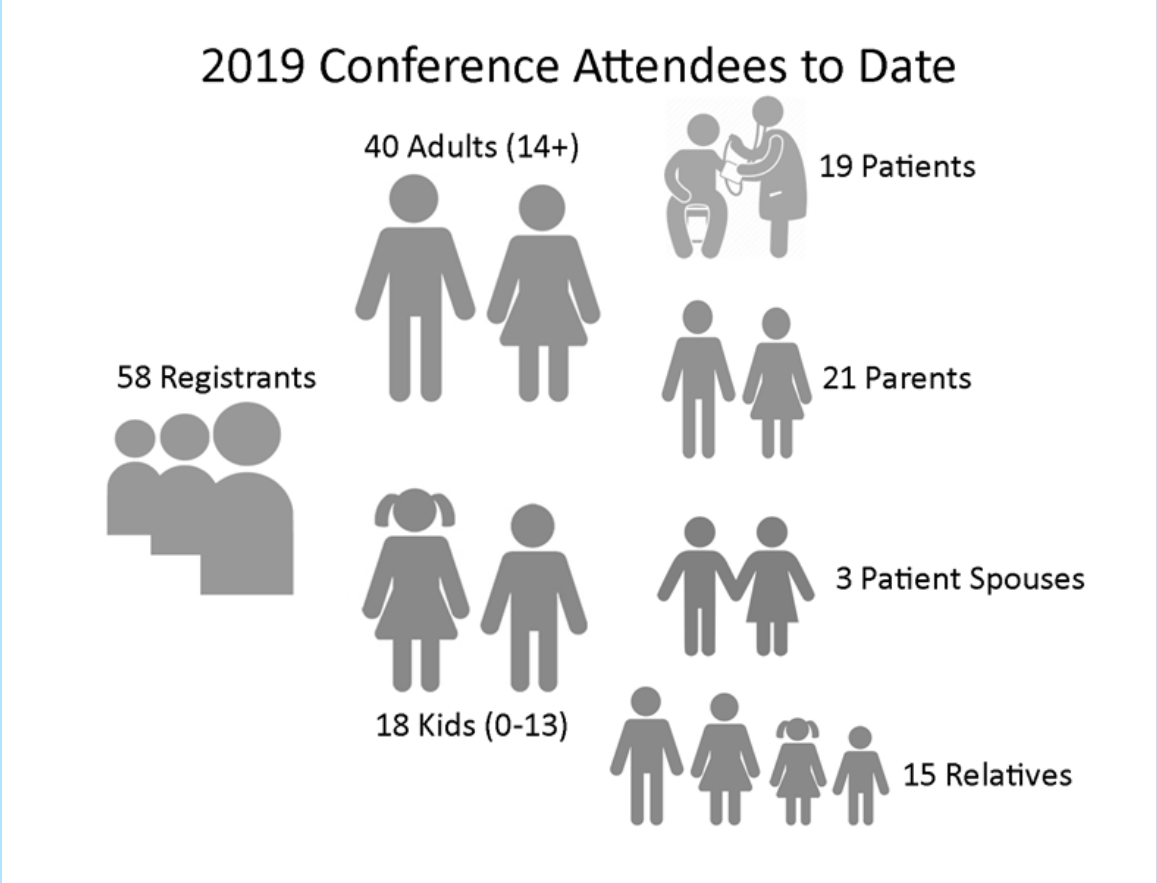
How do my miles translate to money raised?

After a racer is registered, they are set up with their own personal donation page. You can direct those who would like to donate to your race link.

What is the cost and how do I register?

Registration is $14 per participant and all finishers will receive a custom race medal. This would be a great way to involve your family members and friends to support you or your loved one with HCU. To learn about other swag or to register, please visit: https://hcunetworkamerica.org/virtual-race

HCU Network America Conference



Registration is now live. Put the pedal to the metal, register today!
https://hcunetworkamerica.org/2019-conference

Nominations for the HCU Hero Award

We are asking for nominations this year by the HCU community to help HCU Network America choose our HCU Hero Award recipient. The award will be presented at our national conference held October 19-20, 2019 in Indianapolis, IN.

The criteria for the award is that is goes to individuals at the pinnacle or toward the end of their career, recognizing their excellent body of work over a long time to improve the lives of people with HCU. They should have had a significant positive outcome along the way.

Previous awardees have included:
2018: Dr. Harvey Levy

To nominate someone qualified and deserving of the 2019 HCU Hero Award, please send an email to Danae' Bartke, Executive Director, at dbartke@hcunetworkamerica.org with the subject line noting: HCU Hero Award Nomination

Please provide their name, title and a brief summary as to why you feel they are deserving, including work history, volunteerism and other ways they have gone above and beyond for those with HCU.

Deadline is Sunday, August 31st at 11:59 p.m. CST.

ACMG and SIMD Recaps

What is ACMG?

The American College of Medical Genetics and Genomics (ACMG) is an organization that consists of individuals, medical professionals, foundations and corporations who understand the importance of medical genetics, genomics and genetic counseling in healthcare. The organization helps recruit future medical genetic and genomics, develops practice guidelines for physicians, and builds awareness for medical genetics to the general public.

What is SIMD?

The Society for Inherited Metabolic Disorders (SIMD) is an organization composed of clinicians and researchers who strive to maintain and further develop their and others' knowledge of inborn errors of the metabolism (like Homocystinuria). They aim to communicate new discoveries and breakthroughs, while also advocating for advancements in newborn screening and better health reforms.

Conference Topics and Organizational Outreach

The first of these two conferences was the ACMG Annual Genetics Meeting. The conference was held from April 2nd through 6th in Seattle, Washington, with exhibit hours the 3rd through 5th. This was HCU Network America's first time attending the conference and was by far the most diverse conference we have attended to date with also the largest medical professional attendance. The 3 most prominent attendee professions were genetic counselors, clinical geneticists, and laboratory directors and specialists. Like other conferences, the ACMG Meeting had keynote speakers and poster sessions – unlike other conferences we have attended, attendees had the opportunity to attend workshops, Plenary Sessions, Platform Presentations, and Learning Lounges. Between these experiences, exhibits were open for attendees to stop by and learn more. We had wonderful outreach to the diverse group at this conference. Most of those who had stopped by had not heard of HCU Network America; a great reminder of why outreach is so critical!

There was a poster at this meeting sponsored by Orphan Technologies on the incidence of HCU based on medical claims data, showing that in later years there are more patients being treated for HCU than for PKU, presumable due to missed diagnoses at younger age for HCU. There were also many presentations on advances in therapy for genetic diseases, including gene therapy and gene editing, which could someday lead to new therapies for HCU.

On the last day of the conference, ACMG and SIMD had a joint session which focused on metabolic genetics. This joint session was designed to create awareness and education for those who work in the broader field of genetics and to get them think about the possibility of metabolic genetics diagnoses when they see patients. After the joint session was over we packed our things and then headed over to Bellevue, Washington to continue our outreach at the next conference.

ACMG and SIMD Recaps, Cont.

The SIMD annual conference was held from April 7th through 9th in Bellevue, Washington. This was our second time exhibiting at this conference and was the largest SIMD conference to date! Like ACMG, the 3 days were comprised of poster sessions, keynote speakers, panels and a lot of time to network. The topics that were included were Challenges in Organ Transplantation for IEMs, Diagnosis of IEMs: Complementary Use of Genomics and Metabolomics, Improving Patient Care in the 21st Century, and Eye in IEMs. As with ACMG, there were many presentations at this conference on novel therapeutic approaches to diagnose and treat genetic disorders which could someday benefit HCU patients.

During coffee breaks and receptions, the exhibits and booths were open to those in attendance. With each break we saw a lot of new and familiar faces who wanted to learn about the organization and the resources and materials that we have available to both clinics and patients. At this conference we were able to share the many new resources we have developed this past year and years prior. Clinicians were thrilled to know that there are so many resources available to them and the community!

Advancing Homocystinuria Newborn Screening: Presentation to the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)

What is the ACHDNC?

The Advisory Committee on Heritable Disorders in Newborns and Children is established under the Public Health Service Act and is governed under the provisions of the Federal Advisory Committee Act.

"The Committee provides advice, recommendations, and technical information about aspects of heritable disorders and newborn and children screening to the Secretary of Health and Human Services (HHS) for the development of policies and priorities that will enhance the ability of the State and local health agencies to provide for such screening, counseling and health care services for newborns and children having, or at risk for, heritable disorders."

The Committee developed and updates over time the Recommended Uniform Screening Panel (RUSP). "The RUSP is a list of disorders that the Secretary of the Department of Health and Human Services (HHS) recommends for states to screen as part of their state universal newborn screening (NBS) programs. Disorders on the RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments. It is recommended that every newborn be screened for all disorders on the RUSP. Most states screen for the majority of disorders on the RUSP; newer conditions are still in process of adoption. Some states also screen for additional disorders."

HCU Network America's Presentation to the Committee

On Tuesday, April 23, 2019, HCU Network America's Executive Director, Danae' Bartke, President, Margie McGlynn, and parent of HCU patient, Elizabeth Carter presented public comments to the Committee in an effort to convince the Committee to recommend improvements to the newborn screening (NBS) process for HCU in newborns . Danae' Bartke opened with an introduction, including her brief patient story and experience. She then spoke of the inadequacies in current NBS methodologies for HCU and presented anonymized data on HCU patients missed by NBS that the organization has collected from families. She closed her comment section by asking the Committee for their support in revising NBS.

After Danae's presentation, Elizabeth Carter, parent of a young boy with HCU, shared her family's very moving story on the diagnosis of little Elliott. Elizabeth helped paint her family's experience from Elliott going from being a sweet, bubbly toddler on the beach, to fighting for his life after suffering from a series of seizures after multiple blood clots to the brain. She shared the fear she experienced after the doctors said to her "We want you to know how serious this is. We don't expect to lose Elliott, but you need to know that we could." Eleven days after being admitted to the ICU, Elliott received his diagnosis. While there was relief, it also bought more questions. She then closed by sharing that Elliott is now doing well thanks to the various doctors, therapies and diet; in fact she refers to him as "Elliot 2.0". She concluded by asking the Committee to develop improvement to the laboratory test for NBS, so families do not have to relive her family's nightmare.

HCU Network America's President, Margie McGlynn then spoke last. She shared her own experience of the loss of her two sisters due to HCU at a young age. She spoke how far we have come with diet and treatment, and had they been born today they would have had a chance of "living a healthier, longer life". She then presented what we believe is the best solution to enhance current NBS for HCU, a two tier test that would lower the methionine cut off and then use a second tier test to analyze homocysteine and methylmalonic acid. Longer term the organization feels testing Homocysteine as a first tier test would be best, but would take additional research and work. She then wrapped up the organization's public comments by thanking the Committee and offering our assistance in moving the dial forward on HCU NBS. The Committee thanked us for bringing our story forward and gave an assignment to the laboratory standards subcommittee and the CDC to come forward with recommendations for changes to the NBS process for HCU. To view the footage of the presentations, please visit: https://www.youtube.com/watch?v=y6uVQrSBFFc&list=PLvz3MWdu8TP3KY23lZY8ZcK6d95dNyroe&index=2

VMP, LLC
expanding genetic horizons...

Voice: 404.793.7800
Fax: 866.744.5665
www.vmpgenetics.com

HELP US TEACH PHYSICIANS ABOUT HOMOCYSTINURIA

FACT! Teaching about metabolic diseases in medical school and residency programs is poor.
FACT! Most patients live and die without a diagnosis being made, especially when the disease presents in adulthood.
FACT! Patients cannot access effective therapies unless a proper diagnosis is made.
FACT! The sooner a diagnosis is made and treatment begun, the better the outcome.

WE NEED YOUR HELP!

We at VMP Genetics believe in the power of "patient-teaching" and are bringing patients and families into lectures and presentations - at conferences and in the classroom around the country. While doctors teach facts, patients tell stories. Story-telling is a more compelling teaching method with better recall over time than didactic lecturing. We also believe that doctors are more likely to make a diagnosis if they have already seen a patient and heard her/his story. Story-telling can be live or taped...

WE ARE LOOKING FOR...

- ***Patients and/or family members who are interested in telling their stories in local medical classroom settings...*** We are developing a Patient Teacher Registry. If a medical school faculty member is looking to introduce the patient story in a teaching session, the Registry can tell him/her if there are patient-speakers in the area and what diagnoses they have.

- ***Patients and/or family members who are interested in having their stories videotaped...*** As we secure funding, we are interested in recording stories that reflect the broader patient experience. The more variety in the stories, the richer the learning potential.

- ***Videos of patients and families telling their stories...*** A 5-or 10-minute clip can be downloaded into a lecture about that disease or relevant biochemistry to enhance the learning potential of the session.

Please help us in our efforts to raise awareness about Homocystinuria through this innovative educational outreach to the medical community. To Volunteer or participate, or for more information about this project... please contact Jacob Athoe at PatientTeacherRegistry@vmpgenetics.com

Mark Korson, MD
VMP Genetics
Director of Education

Jacob Athoe
Genetic Counseling Student
Boston University Genetic Counseling Program

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/

We'd like to thank the following content contributors:

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