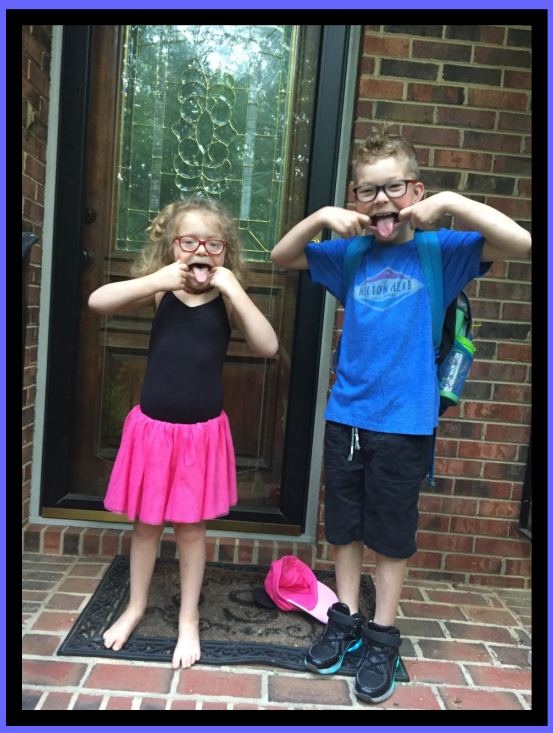
HCU Herald

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:
Clara and Jacob from North Carolina

The journey to diagnosis was most definitely not an easy one. Jacob (6 at the time) and Clara (4 at the time) were both diagnosed in 2018. The beginning of our journey started when Clara started struggling to see. When she was about to turn 3 we noticed she was holding objects very close to her face, so we took her to see our optometrist. We were informed that Clara was extremely nearsighted, -10 myopic. We thought it was odd as no one else in our family has vision trouble. After about 6 months we noticed her struggling to see again, while she was wearing her glasses. Back to the optometrist we went, this time her eyes read -18! Being concerned that there was something more severe going on I asked the optometrist what she thought it could be. She referred us to a pediatric ophthalmologist. Upon further investigation by the ophthalmologist, Clara was diagnosed with ectopic lentis or subluxation of the lenses. The doctor said there was a high probability it was caused by a genetic defect, and we should consider seeing a geneticist.

During this time her pediatrician suggested we test for food allergies as well as other possible causes to her drastic eye changes, such as autoimmune disease, inflammation, etc.; all of which came back abnormal. The test showed food sensitivities to eggs, dairy and gluten, but otherwise gave us no answers. Then the plethora of doctor appointments came: rheumatologists, hematologists, neuro-opthaologist, more and more tests, and blood work; but still no answers.

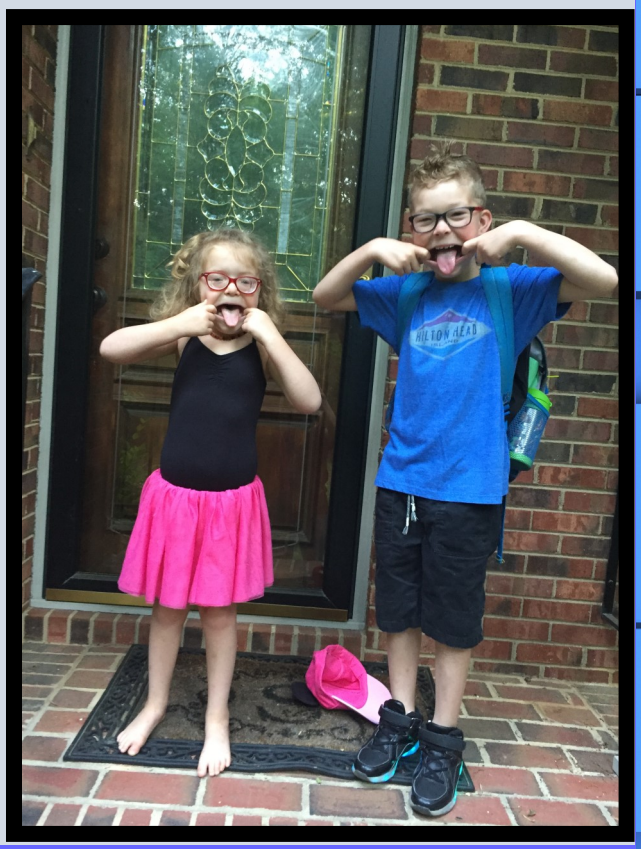
Then came a day where Clara began to vomit, and wouldn't stop. The doctor was certain it was a stomach virus that would pass in 24 hours. 24 hours came and went and she continued to be very ill. She had become dehydrated and she was refusing fluids. Then she vomited bright green and I decided it was time to go to the hospital. After some tests we discovered she was having an attack of pancreatitis. Pancreatitis is exceptionally rare in children. The doctors said it was probably a byproduct of a virus, or she could be predisposed to pancreatic issues.

The following week we had an appointment with a rheumatologist. She looked at her medical history and labs. She was confident that she didn't have an autoimmune disease. She did a Google search, that's right, a Google search for a correlation between ectopic lentis and pancreatitis. She examined her previous labs again and told us she was quite certain that Clara had Homocystinuria. She told us a geneticist would be contacting us.

Within a few weeks we saw a geneticist who confirmed her diagnosis. We then began seeing a team of physicians and dietitians at UNC Chapel Hill. 5 months later the lens in her right eye became partially detached and was pushing forward through her iris. She had surgery to remove the lens at Duke University. She will be having the other lens removed as well.

Jacob and our other daughter, Aubrey were also tested for HCU. Aubrey tested negative. Jacob, however, tested positive. Jacob has always struggled with meeting most physical milestones (fine and gross motor, as well as speech). He would make progress and often times backslide. We never really understood why, until his diagnosis. We have definitely seen a lot of progress in these areas in just a few months post diagnosis. It is disappointing that HCU wasn't caught during their newborn screens. Clara's was actually a borderline high read for methionine, unfortunately we were never informed or told a retest should have been done.

Both children are doing great adjusting to their low protein diets. As a family we support and encourage them as much as we can. We are showing them how to make smart choices while dining out or at family/friends gatherings. We are also teaching them how to read labels on snacks/food. They both take Cystadane without any struggle (anymore). We mix it with either chocolate almond milk or lemonade. The formula is not an issue with Clara. We have found that she likes the HCU Cooler with the red lid, mixed with cherry limeade powder (TruLime). Sometimes Jacob tolerates the formula well, sometimes he has a difficult time getting it down. He usually does the HCU Express mixed with vanilla coconut milk, chocolate syrup, vanilla syrup, and peppermint extract.



Clara and Jacob encourage and support each other during doctors' appointments, blood work, tasting new formulas, trying new foods, etc. It has been very sweet watching their bond grow through HCU. As much as we were sad when we found out Jacob had the disorder, we were able to find the silver lining, knowing that Clara isn't alone in her diagnosis. When we told her he had HCU, her eyes lit up. My husband, Jeremy, found them in a full embrace. She later thanked Jesus that Jacob had homocystinuria. She loves having that connection with him.

HCU and You: Connecting the Dots

Connecting the Dots is an ongoing series of articles meant for individuals affected by homocystinuria (HCU) and their families. In previous articles, I've written about day-to-day questions and concerns families often bring up. I also want to review relevant research as it is published as part of this ongoing series of articles. In January 2019 the journal Genetics in Medicine published an article by Mohamed A. Almuqbil, M.D., and his colleagues on common mental health problems experienced by people with HCU.

Data were collected through a review of medical records. Newborn screening identified 14 of the 25 people with HCU. All but one of the people with HCU were on a combined treatment that included two or more diet, betaine, B_6, Folic acid, B_{12}, and cysteine. Anxiety and depression were the most commonly reported mental health difficulties, which is similar to patterns of data for the general population in the United States. Other difficulties reported by people with HCU included anger, impaired attention, and hallucinations. The presence of anxiety or depression was most common among people who had HCU and impaired intellectual functioning. The chance of mental health difficulties was found to be higher with an older age at time of diagnosis.

These recently reported findings highlight the importance of access to mental health evaluations and treatment for people with HCU, and the availability of mental health services is particularly important for people who have HCU and impaired intellectual functioning. For families, this study highlights the importance of understanding the HCU can impact a wide range of areas including emotions and behaviors.

HCU was first described in the 1960s, but for a variety of reasons, documenting the impact of HCU on day-to-day emotions and behaviors has been difficult. The barriers to fully understanding the possible impact of HCU includes the rarity of the diagnosis, the wide variability in severity and presentation, and the large number of people who are missed during newborn screening. The exciting news is that interest in studying the daily impact of HCU is growing, and a number of potential treatments are being developed. In October 2018 HCU Network America held a webinar "2018 Global Research Map Update" that provided an overview of potential treatments based on a project they co-sponsored with HCU Network Australia.

I hope to continue to include reviews of relevant research as part of the Connecting the Dots series. The more we know about HCU, the more families can be prepared for challenges and medical teams can make informed treatment choices. If you have ideas for future articles, you can email info@hcunetworkamerica.org

Sincerely,
Ben

Benjamin D Goodlett, PhD, is a psychologist who specializes in working with children and families affected by inborn errors of metabolism.

To access the publication this article references, please click here

Recipes from the Kitchen

Braised Cabbage

Author: Amber Gibson

Serves about 7 - 3 oz. servings.

Protein: 1.1 g per 3 oz. serving

Calories: 75

Prep time: 15 min | Cook time: 30 min | Total Time: 45 min

Ingredients

- 1 TBSP Bacon fat drippings (fat only, strained)
- 80 g Diced Onion
- 2 clove(s) Minced Garlic
- 432 g Cabbage, red, raw, shredded, about one small red cabbage
- 2 TBSP Packed Brown Sugar
- 1/3 c White Wine
- 1/4 c Apple Cider Vinegar
- 1 c Vegetable Broth
- 100 g Peeled, Diced Apples

Directions

1.

In a large sauce pan heat the bacon fat over medium low heat. Once hot, add the onion and saute until translucent and fragrant. Add the garlic and sauté for about a minute, making sure to stir constantly. Add the shredded red cabbage and brown sugar. Stir until covered with the bacon fat and sugar dissolves. I like to use tongs to do this.

2.

Add the white wine to the cabbage and stir. Continue to cook for about 3 minutes, stirring occasionally. Next add the apple cider vinegar and vegetable broth. Cook over medium low heat until the cabbage has wilted, but still a little firm to the bite. This will take about 25 minutes. Add the apples last and cook for an additional 5 minutes. Remove from heat, season with salt and pepper to taste. Serve warm.

Recipes from the Kitchen

Irish Colcannon

Author: Amber Gibson

Serves about 10.5 - 1/3 cup servings.

Protein: 2 g per 1/3 cup serving

Calories: 148

Ingredients

- 700 g Chopped White Potatoes
- 1 1/2 tsp Cambrooke Chicken-Flavored Consommé & Seasoning, dry, divided
- 2 c Water
- 175 g Parsnips, raw, slices,
- 8 TBSP Butter, regular or unsalted, cut into pieces
- 40 g Diced Onions
- 280 g Shredded Green Cabbage
- 1 tsp Dried Chives
- 1/2 c Rice Dream, Original

Directions

1. In a medium pot add the potatoes and 2 cups of water and one tsp of Cambrooke's consommé powder. Cook over medium heat until the potatoes are for tender. Drain over a bowl and reserve the potato water. We will use this again later! While potatoes cook, steam the parsnips.

2. In a large rimmed skillet add the butter and melt over medium low to medium heat. You do not want the heat too high as you do not want the butter to burn. Cook the butter until it has a nice nutty aroma and has browned some. Remove 1/4 cup of the butter and set aside for use later. Add the onions to the skillet and sauté over medium heat for one minute. Now add the cabbage and chives. Gently coat in the browned butter and onions. I suggest using tongs to do this as they make it easier to turn the cabbage. Continue to cook the cabbage until wilted and begins to brown.

3. While the cabbage cooks, mash the potatoes and parsnips together. Add the rice and mix to combine. Now you can add the reserved potato broth a little at a time until the mashed vegetables are nice a fluffy. It took about 1/4 cup of the broth for me. I saved the rest for another use.

4. Remove the cooked cabbage from the skillet and add to the mashed vegetables and gently fold to combine. In the skillet add the remaining browned butter we set aside from earlier. Add the rest of the potato broth and cook until the sauce reduces by a third, about another three minutes or so. You can cook a little longer if too thin.

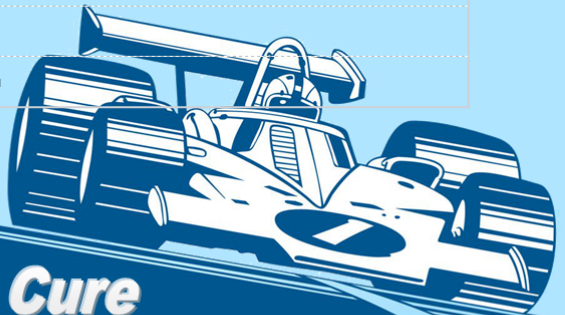
5. Add the butter sauce to the colcannon and stir to combine. Serve hot. Enjoy

*Note: Be cautious when seasoning with added salt and pepper. The broth can be salty, which seasons nicely on its own.

HCU Network America Conference

Agenda At A Glance

Saturday	Topic
7:30—9:00 am	Registration and Vendors Open
7:45—8:45 am	Breakfast
9:00—9:45 am	Introduction and Meeting Expectations
9:45—10:45 am	Keynote 1: Natural History Study Update
10:45—11:15 am	Vendor Break
11:15—12:00 pm	Keynote 2: Best Practices in Treatment of HCU
12:00—1:15 pm	Lunch
1:15—2:15 pm	Keynote 3: Screening Family Members and Family Planning
2:15—2:45 pm	Vendor Break
2:45—4:00 pm	Breakout Sessions: By age group
4:00—5:30 pm	Free Time
5:30—8:30 pm	Reception (Dinner Included)
Sunday	**Topic**
7:30—8:30 am	Breakfast, Registration and Vendors Open
8:45—8:55 am	Group Photo
9:00—9:30 am	Vendor Acknowledgement and HCU Hero Award
9:30—10:30 am	Keynote 4: Therapies on the Horizon
10:30—11:00 am	Vendor Break
11:00—12:00 pm	Panel: Ask the Expert
12: 00—1:30 pm	Closing Remarks and Lunch

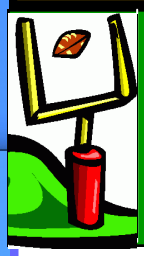


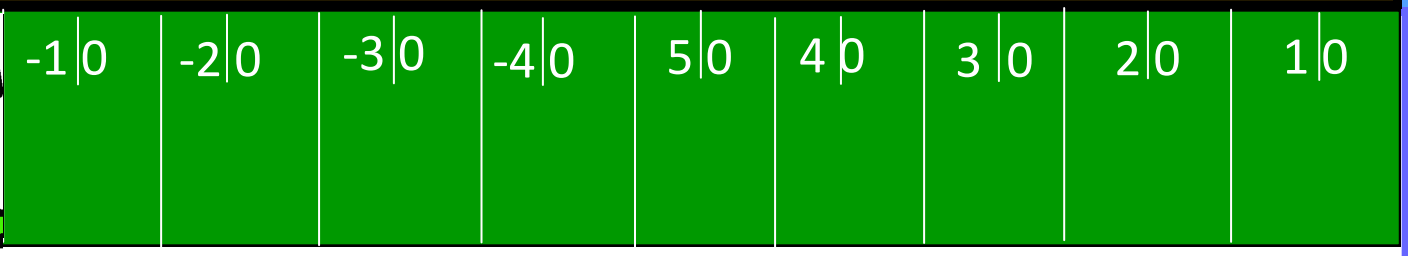
*Schedule is subject to change.

Accelerating Towards a Cure
2nd Homocystinuria Conference
October 19 & 20, 2019 | Indianapolis, Indiana

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

Fundraising Idea: Sporting Events Tournaments





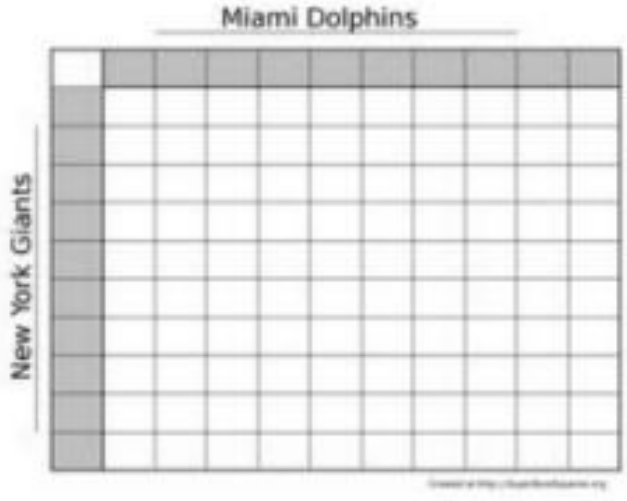
What do NFL football and college basketball both have in common, other than they are both sports? Well, they both center around a final end game. For Football, the final NFL game is the Super-Bowl, for College Basketball, the final event happens during March Madness.

These big sporting events are a great way to not just show your team spirit, but they are a fantastic way to raise funds for your charity of choice—HCU Network America!

There are two most common types of fundraisers around these two are Super-Bowl Squares and March Madness Brackets.

Squares Fundraiser:
How it works: The cost is $5 or $10 donation per square. The purchaser writes his or her name in the square, and then the seller (or someone not playing the game) will randomly pick numbers from 0-9 for each team in the game, and assign that number to a particular row or column (the grey shaded columns below). These numbers represent the last number in the score of each team at the end of the game. In other words, if the final score is Lions 17 - Saints 14, then the winning square is the one with a Lions number of 7, and a Saints number of 4. Since no one knows what numbers each square will represent, the odds are the same for everyone. So good luck and enjoy.

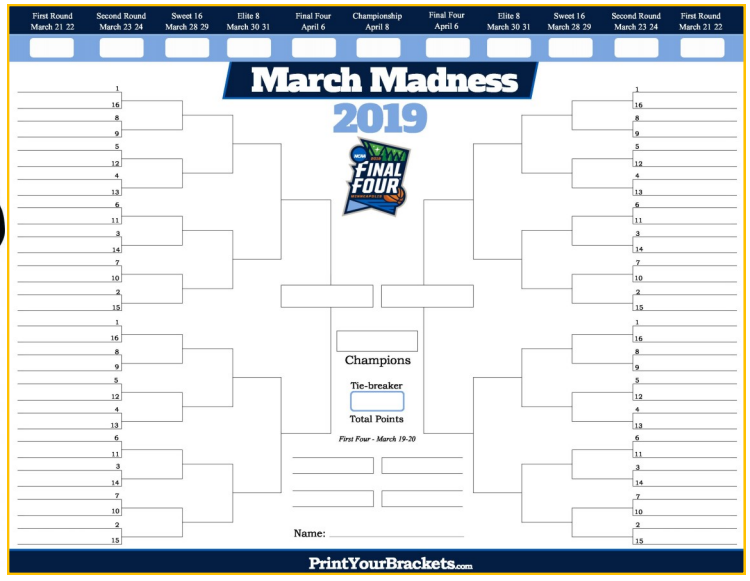
Winner Breakdown: If you sell 100 squares (must sell all 100 squares to host the fundraiser) at $10 each – the funds raised are $1,000. You can chose to split the pot evenly and the winner will get $500 or get a large item donated for the winner and keep all funds raised toward your fundraising effort. (You can also decide to split the prize money and also give some at halftime or the end of each quarter).



For further instructions and a printable template visit:
https://www.printyourbrackets.com/nflweekly100squares.html

For online tools check out:
http://footballsquaresonline.com/
https://www.runyourpool.com/nfl-football-pools.cfm

Fundraising Idea: Sporting Events Tournaments



March Madness is a three week period packed full of buzzer beaters and is a sports poolers dream. But how can you keep the excitement alive when most of your members have had their brackets busted? A great option is our Madness Squares pool format, as every game of the tournament will have a winner!

How Do Madness Squares Work?

If you are familiar with Super Bowl Squares, the main idea is the same for March Madness. A 10x10 grid of boxes is setup and each row and column are given a number from 0 to 9. Just like in Super Bowl Squares, each square of the grid can be claimed by a pool member.

Winner breakdown:

Each round is worth a set number of points, you can determine this on your own, but be sure to let all of the entries know before the tournament begins what the scoring system will be (you should write the point values under each round at the top of the bracket).

Declaring a Winner

Multiply the total number of correctly picked games in each round by the points assigned to that particular round. Tally all rounds together and the person with the highest point total wins!

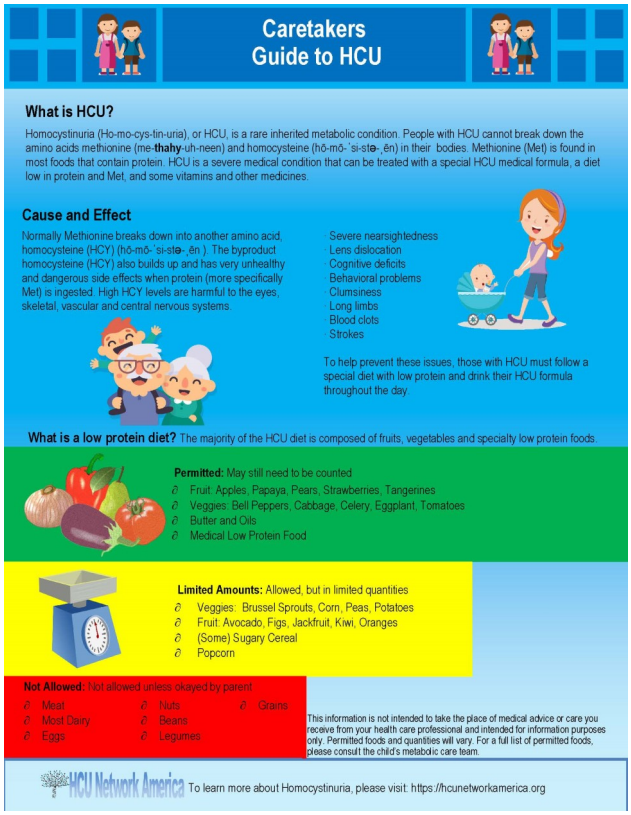
For further instructions and to print your bracket, visit:
https://www.printyourbrackets.com/howtomarchmadness.html

For online tools, check out:
https://www.runyourpool.com/march-madness-squares-pools.cfm

Caretakers' to HCU

Inevitably you will have to leave your child who has HCU with someone – whether it's for work, to run errands, a doctor's appointment, vacation, or to simply give yourself a break (let's be real – all parents need a break once in a while); you will eventually have to leave them with a babysitter, relative, or grandparent.

For many parents the first time you leave your child with HCU under the care of someone else, it will likely cause a lot of stress and anxiety. Try to breathe and remember, you are not the first parent who has gone through this! To help support you in this new chapter of raising a child with HCU we have developed a Caretakers Guide for HCU. Our Caretakers Guide to HCU will help you explain what Homocystinuria is, what foods are allowed, and how your child's caretaker can help make this transition as smooth as possible for you and your child.



To download and print the Caretakers' Guide to HCU
Visit: https://hcunetworkamerica.org/toolkits-and-checklist/

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:

Editor in Chief: Danae' Bartke
Heroes of HCU: Clara and Jacob from North Carolina
HCU and You: *Recipes from the Kitchen: Amber Gibson*
HCU and You: *Connecting the Dots: Benjamin D. Goodlett*
Caretakers Guide to HCU: Patient—Parent Advisory Committee
Fundraising Tip: Fundraising Committee

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