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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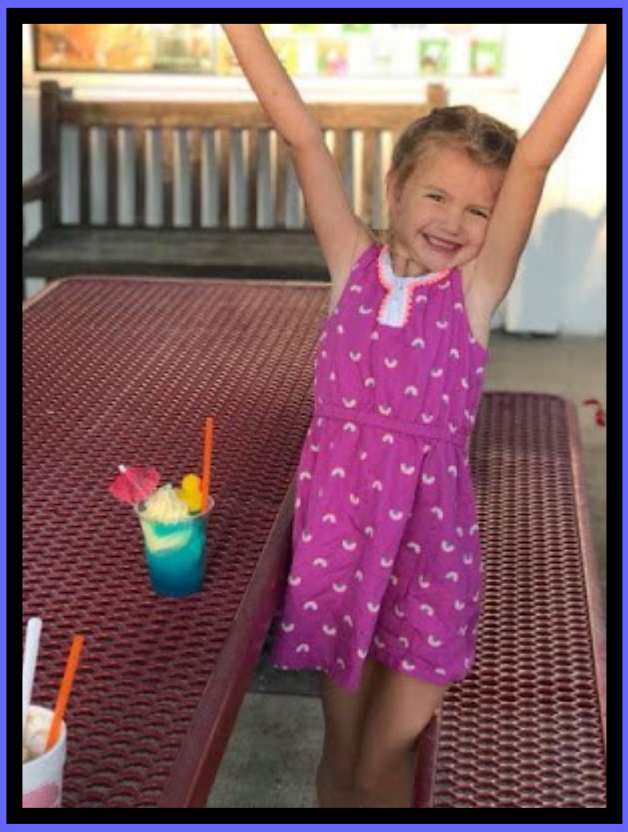

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POA Advertisement

New and innovative product -for the dietary management of HCU

Now available in the US

Request a sample today

EASY HCU® Tablets

HCU Easy Tablets is a methionine free protein substitute tablet containing essential and non essential amino acids, to be used in the dietary management of patients with HCU (homocystinuria) aged 3 years and above.

HCU Easy Tablets are coated (slow release) tablets which are to be swallowed with liquid.

Patients love the tablets for many reasons.....

- Easy to take
- No taste
- No odor of amino acids on the breath
- Less stomach problems
- Easy to travel with
- No mixing required
- 10 grams of protein = only 11 tablets

Request a free sample at order@poapharmana.com

poa Pharma North America

www.poapharma.com

HCU Hero:
Ella from Michigan

The greatest blessing I received was in September 2012 when I became a mom for the first time! Ella, our first child, was born a healthy and beautiful baby girl. Holding her in my arms and looking into her eyes, I was immediately in love with her. My husband and I took her home from the hospital at one day old. Then, when she was only 4 days old, we received a phone call saying her newborn screening came back abnormal. We were told it was most likely a false positive because the condition is very rare. We took Ella in for more blood work and quickly confirmed that she does have a rare disorder called Homocystinuria.

Ella is now six years old and doing absolutely fantastic. She is healthy because she eats a very restrictive diet of only 9-10 grams of protein from food per day. She drinks Hominex 2 twice per day, and takes vitamins along with Betaine to help her break down the protein in her body.

Ella is a smart, caring, creative, and goal driven little girl! She is learning to read nutrition labels and count the grams of protein she eats daily. The hardest parts of having Homocystinuria are getting her blood work drawn on a regular basis, knowing there isn't a cure, and telling her that no, she can't eat certain foods other kids are eating.

We are thankful for newborn screening, thankful for a wonderful metabolic doctor here in Michigan, thankful for those that raise awareness and funds for HCU research, and thankful there are low protein foods and formula that are available for Ella.

We tell Ella that God made her so special when she was created!

Recipes from the Kitchen

Low Protein Deviled Eggs

Author: Amber Gibson

Servings: 12 eggs
Protein: 0.28 g per egg
Calories: 29 per egg

NOTE: Filling is not part of the overall protein

Ingredients:

- I c. Coconut Milk, Canned
- 2 tsp. Agar Powder
- 1/2 tsp Salt, Table

Directions:

1. Combine coconut milk, agar agar powder, and salt in a small sauce pan and use a whisk to combine. Bring to a slow boil over medium heat.

2. Remove from heat and pour into egg molds. I used an egg tray from the dollar store and a syringe to fill the cavities. Refrigerate for 30 minutes to set. Fill with your desired filling.

Fillings for center:

- Guacamole
- Mashed potatoes mixed with traditional deviled egg spices (mayo, vinegar, mustard, salt and pepper).
- Low protein potato salad (see cookforlove.org for recipe)
- Alouette spreads mixed with mayo to soften

Recipes from the Kitchen

French Toast Bake

Author: Amber Gibson

Servings: 4 Servings
Protein: 0.3 g per serving
Calories: 232 per serving

Ingredients:

- 4 slice(s) Cambrooke Cinnamon Raisin Swirl Bread, cut into cubes
- 30 g Vanilla Pudding, Instant, dry mix only
- 1 TBSP Egg Replacer, I used Ener-g Egg Replacer
- 2 TBSP Sugar, Brown, packed
- 1/8 tsp Salt, Table
- 1/4 tsp Cinnamon, ground
- 1 c Rice Dream, Original

Directions:

1. Preheat oven to 350 degrees. Spray a 1.5 to 2-quart casserole dish with nonstick cooking spray and set aside.

2. Place bread cubes in a small bowl and set aside. In a 2-cup liquid measuring cup, add the rice milk, pudding mix, egg replacer, brown sugar, salt, and cinnamon. Lightly whisk to combine. Pour over the bread cubes and gently toss until all cubes are coated. Pour into prepared casserole dish. Bake for 25 to 30 minutes. The custard mixture will be set. Serve hot with maple syrup.

Recipes from the Kitchen

Crepes

Author: Amber Gibson

I created these looking for a similar match to the palachintas (Hungarian pancakes or crepes) we make in my family. These work perfectly and can be filled with anything from cookie butter, jelly or jams, fruit, chocolate ships, etc.

Servings: 14 crepes
Protein: 0. 1 g per crepe
Calories: 54 per crepe

Ingredients:

- 100 g Cambrooke MIxQuick Baking Mix, Gently packed
- 8 g Egg Replacer
- 235 g Rice Milk, Divided 21mg
- 28 g Butter, regular or unsalted, melted
- 1/4 tsp Salt, Table
- 1/2 tsp Vanilla Extract

Directions:

1. In a small bowl mix the one tablespoon of egg replacer with 1/4 cup of the rice milk. Set aside to sit.

2. In a medium bowl add the mixquick and salt. Gently whisk to combine. In a 1-cup measuring cup add 3/4 cup of rice milk, the melted butter, and the vanilla. Stir to combine. If butter solidifies, microwave for 15 seconds and stir. Add the rice milk mixture to the dry ingredients and mix to combine. Add in the egg replacer mixture and mix. Batter should be a little thinner than pancake batter.

3. In a small skillet over medium heat, melt 1 teaspoon of butter. Once heated add 3 tablespoons of the batter to the skillet. Swirl the skillet to spread the batter around in the pan. You want a very thin layer of batter. Cook the crepe until lightly browned, about 3 to 4 minutes. Flip crepe and cook 30 seconds to set the batter on the other side. Remove from pan. Continue this step for the rest of the crepes. Serve immediately with you choice of fillings and toppings.

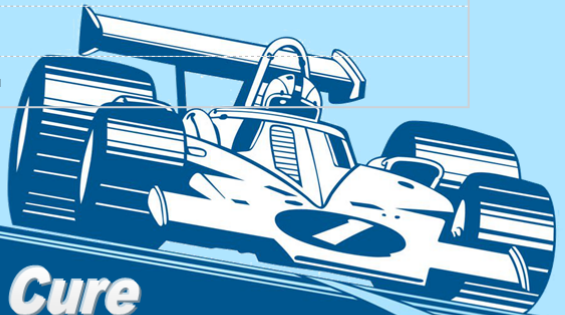
Notes:

These can be frozen and reheated if necessary.
I have a crepe maker by Cucina Pro that I adore that I use for these crepes. It makes it easy to measure and use.
If the crepes are cooking too fast, turn heat down to medium low.

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

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. To learn about other swag or to register, please visit: https://hcunetworkamerica.org/virtual-race

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!

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CLICK HERE TO REGISTER

MEDICAL FOOD INSURANCE COVERAGE TIPS FOR ALL TYPES OF INHERITED METABOLIC DISEASES AND RARE GENETIC DISORDERS IN THE USA

-Raenette Franco

The complexity of the health care system can overwhelm even the savviest patient and/or clinical professional. That is why Compassion Works Medical was created to hold hands with patients and alleviate the clinics' time through the difficult process of medical food insurance coverage.

Compassion Works Medical was founded by Raenette Franco, CEO and Certified Billing and Insurance Specialist. Raenette has been inspired to share her hands-on experience with medical foods insurance coverage specialized in all types of inherited metabolic diseases and rare genetic disorders. Compassion Works Medical works with you and for you, providing guidance and supporting you with compassion and integrity. Ms. Franco specializes only in medical food coverage and has been battling insurance coverage for medical foods for over a few years in addition to fighting for the Medical Nutrition Equity Act on Washington's' DC Capitol Hill. Compassion Works Medical collaborates with patients' current insurance providers, and fights for state mandated coverage.

Every case is unique and different. Understanding your options and insurance terminology is essential to obtaining the coverage that you deserve!

Let's start with the basics……

What is a Medical Food? You may hear these words often and could be confusing to the words "formula" or "dietary supplements".

- Medical foods are foods that are specially formulated and processed (as opposed to a naturally occurring foodstuff used in a natural state) for a patient who requires use of the product as a major component of a disease or condition's specific dietary management (i.e. designed for a certain disease) and intended to be used under medical supervision.

- Formula is basically the same thing as a medical food as they are made from the building blocks of foods.

- Dietary supplements are not generally designed for a certain disease, but are used in contribution to maintain health with a disease such as added vitamins. Dietary supplements are sometimes added to patient's dietary management.

- What is Enteral? Enteral is a medical term used for a feeding method either oral or tube feeding; Hence Enteral formula.

Overall Medical foods, dietary supplements and enteral formula are common words uses for insurance coverage.

Coverage for medical foods and dietary supplements is generally under medical benefits and supplied by a durable medical equipment (DME) distributor by using the description of coverage. However, medical foods are also covered under pharmacy benefits by product only, just a little tougher to maneuver insurance coverage.

Coverage for special injectable vitamins: Are you or your family member on injection vitamins such as cyanocobalamin/ hydroxocobalamin ML (B-12)? If yes, coverage for this special vitamin could be challenging. This special injectable vitamin may or may not be covered under pharmacy benefits. Some pharmacy benefits donot cover these products and they are considered over the counter. To obtain coverage, it requires jumping through loop holes. However, the health insurance benefits for cyanocobalamin/hydroxocobalamin ML could be covered under your medical benefits. Since the skin is pinched or broken such as with an injection and the place of service is at your clinic. Depending on the dosage your physician uses, a compounding pharmacy may need to make the right concentration for you. Make sure the pharmacy dispenses the exact product your doctor prescribes. Many patients need hydroxocobalamin, and the pharmacy may substitute cyanocobalamin without telling you so make sure you ask. If your clinic can administer the vitamin and bill your insurance company, it would be the best affordable route.

The insurance language under medical benefits for Hydroxocobalamin ML use HCPCS code J3420 and for Pharmacy benefits it is under an NDC number (i.e. 00591-2888-30). The place of service is at the office – Usually injected at the physician's office under medical benefits.

Insurance coverage tips……

To start investigating coverage for your dietary management, it is recommended to start with your medical benefits first. Sometimes when we get a prescription we automatically think it's a pharmacy benefit and that's natural, however, if you have a prescription for medical foods or dietary supplements it's best to check with your medical benefits first.

Here are eight tips below to better understand your medical food and dietary supplement insurance coverage:

Rule No. 1: Never take NO for an answer!

1. **Learn Insurance Terminology**
2. **Understand the difference between Medical and Pharmacy coverage**
3. **Resolve reimbursement issues between insurance company and supplier**
4. **Verify Insurance Benefits before placing an order**
5. **Look out for Medical Food Exclusion Removal**
6. **Understand Gap Exceptions for policies with no out-of-network benefits**
7. **Be prepared for a prior authorization before covered services**
8. **Got Medicare or Medicaid? Best Avenue for medical food coverage.**

It's important to understand your health plan's guidelines for medical food coverage by thoroughly reading your health plans summary of benefits to find out if your medical foods and dietary supplements are covered. Start by looking under durable medical equipment benefits (DME) and non- covered services including exclusions. Key words: **ENTERAL, MEDICAL FOODS, NUTRITION, FORMULA, SUPPLEMENTS.**

Insurance Terminology: Medical food and dietary supplements coverage is a complete foreign language to the health insurance industry. There are certain terminologies used to help obtain the most accurate coverage details with your health plan's benefit specialist such as;

- Service codes (also known as HCPCS codes) used to describe the medical foods, enteral formula, dietary supplements and vitamins (i.e. B4155, B4157, B4162, B9998, S9435, S9435, J3420). These codes could be administered orally, tube feeding or vitamin injection. Injections are usually done at the clinic and not at home for proper coverage.

- In-network and out-of-network to help determine the most affordable way to obtain your dietary needs. Also known as participating or non-participating.

- Know the difference between prior authorization and predetermination. Prior authorization is required before coverage and predetermination is not required before coverage, but helps avoid any future denials.

- Diagnosis driven plan: This is a plan that will only cover if the diagnosis code such as your medical condition(s) matches the description of service. Your diagnosis codes starts with a letter (i.e. ICD-10: E71.121). If it matches then you are covered. Diagnosis driven plans are easily mistaken as not covered, so if your benefit specialist mentions that it's not covered ask if your plan is diagnosis driven.

- Other words are exclusions, out-of-pocket, state mandated plans, deductibles, fully insured, self-funded, allowed amounts, suppliers, gap exceptions.

2. **Difference between Medical and Pharmacy coverage:** Typically medical foods and dietary supplements are generally covered under your medical benefits and provided by a DME distributor. The medical benefits cover these items by using the service codes and the diagnosis code. Pharmacy benefits cover by the product alone and not the service. Medical foods and dietary supplements could be challenging for coverage under pharmacy benefits; if the product is not listed in their system and considered over the counter it's not covered. If you pharmacy plan does not cover your product, then use you medical benefits or file for an appeal.

3. **Reimbursement-Billing issues between insurance company and supplier:** Receiving bills from your providers could be scary. Don't panic! First make-sure you if you've received a bill from your provider or is it an Explanation of Benefits- EOB statement (not a bill) from your insurance company. Check your EOB and match it with your invoice to determine if the bill is for your deductible or co-insurance. If not, contact your provider immediately and go over your invoices. Look out for any unnecessary charges.

4. **Verifying Insurance Benefits before placing an order:** The best way to avoid delays with your medical food orders are to try to verify your benefits and coverage "first" with your insurance carrier before placing an order. Contact your insurance carrier and ask for benefit coverage for Medical Food/Enteral Formula or nutritional supplements. Include diagnosis code (ICD-10). Mention it's "usually covered under DME". Remember to ask if the plan is diagnosis driven to avoid any misunderstandings.

5. **Ask for any exclusion on your policy for medical foods;** if there are any exclusions that means your medical foods are not covered, it is not the insurance company that you would fight with. This is out of their hands. You will have to go to you employer's HR department (assuming you get your insurance through your employer) and ask for a medical food exclusion removal and present your letter of medical necessity explaining your rare genetic disease. Medical food removal template letters can be found at Compassion Works Medical. **To make a request, email raenettef@compassionworksmrs.com.**

6. **What is a Gap Exception**? A coverage gap exception is a waiver from a healthcare insurance company that allows a customer to receive medical services from an out of network provider at an in network rate. Usually HMO or EPO plans do not have out-of-network benefits, but if you can't find an in-network provider to supply your medical foods, you could ask your insurance carrier for a gap exception. Also, your out-of-network supplier could request a Gap exception to your insurance company directly. This way is easier!

    - One of the best reasons for your waiver is that there aren't any other in-network providers within 100 miles of your residence that can supply your medical foods. Another is that you prefer to use the out-of-network provider because of a strong long-lasting and trusting previous relationship.

    - Any request is worth a shot! This also saves time for your out-of-network supplier as well as providing faster medical food service.

7. **Be prepared for a prior authorization that is required by your insurance carrier**: some policies require prior authorizations from your insurance company before they will cover. Prior authorizations need to be identified as "medical necessary". This is usually provided by your clinic or medical food supplier. Make sure your clinic provides you a letter of medical necessity (LMN) with a prescription and recent progress notes (A.K.A. clinical notes). Your provider usually makes these requests.

    - Stay on top of your prior authorization approvals. When they expire, you or your provider will have to request a renewal. This depends on your policy; i.e. month to month, every 3 months, yearly, etc.

8. **Do you have a government plan such as Medicaid or Medicare?** As most of you already know, Medicaid usually follows all of the state mandated laws and covers 100% for in-network providers and may require a prior authorization. Medicare straight from your state does not cover medical foods "UNLESS fed by a feeding tube and is the sole source of nutrition".

- **Want Medicare Coverage?:** If you have already have Medicare you can switch to a Managed Medicare Plan in your state such as AARP, UHC, BCBS, Humana, etc. and they could provide your medical food coverage. You may not have to pay any extra premiums. Many patients are able to be covered through their managed Medicare plans.

This is NOT a Supplement Plan; it is a Managed plan that has leniency for medical food coverage. Supplemental plans only follow the Medicare straight state plans. Supplemental plans will have the word “supplement” on your card or “Complete” .Make sure that your plan is not a supplement as they follow Medicare guidelines. To find a managed Medicare plan in your state by visiting https://www.medicare.gov/find-a-plan/questions/home.aspx Or simply call member services listed on the back of your Medicare card for assistance.

An extra bonus tip * From my experience if you are looking for a reliable dietary supplement, medical food and vitamin supplier for all types of rare genetic disorders, I suggest checking out **Solace Nutrition, LLC**. https://www.solacenutrition.com/ . They are very unique and provide most dietary supplements that you cannot find anywhere else. Ask your provider to search their website for the most appropriate supplement that could help you and your family member’s dietary management. Also, most of their products are covered by some insurance companies.

In addition: If you have homocystinuria, I suggest checking out this unique European company POA Pharma North America http://www.poapharma.com/en/ .Their products are also very distinctive comparing to other marketed in the USA. Also, most of their products are covered by some insurance companies.

I have experienced success with patients affording products from Solace Nutrition, LLC and POA Pharma North America.

The tips above are based on actual experiences. I believe there are no true experts with all of the answers.

So let’s face the facts, patients NEED an Advocate, preferably someone with a medical food- insurance background. Patients need champions who can: (1) TRANSLATE what’s being told (2) ASK THE RIGHT QUESTIONS that patients ‘don’t know to ask’ (3) COMMUNICATE upwards, downwards and sideways.

For support and questions on medical food insurance coverage for all types of Inherited Metabolic Diseases and rare genetic diseases, please contact Compassion Works Medical at (973) 832-4736; email ranettef@compassionworksmrs.com.You will note be charged for this service as I have an agreement to provide with HCU Network American to provide this support , which I am doing pro-bono this year. *Please also join in the webinar listed below*

Still have questions? Want more information about navigating insurance?
Register for our insurance webinar: https://hcunetworkamerica.org/insurance-webinar/

Running for Rare With Chris Hummel

Chris Hummel and his family have been long-time supporters of the HCU community, having raised thousands of dollars towards Homocystinuria research. Chris is at it again. On April 15, 2019, he'll be running the 123rd Boston Marathon and has set a large fundraising goal of $12,000 that will benefit HCU Network America!

What inspired you to run the Boston Marathon?

There are many reasons Chris is motivated to run the Boston Marathon, including honoring the victims of the 2013 terrorist bombings. Chris and his family were at the Boston Marathon that year as spectators. They made the trip to support one of our board members, Kristin Rapp, who was running the marathon that year as part of the Running for Rare Team. Kristin was running in honor of their son, Will. The Hummel family returned to Boston the following year to support Kristin again, and Chris became further involved with the Running for Rare Team.

The Boston Marathon is the oldest, most prestigious marathon in the world. Although spectating was great, Chris has been drawn to run it ever since that first experience in 2013. Chris has run three marathons previously, including the 2015 Providence Marathon which he ran as a member of the Running for Rare Team. It was there that Chris set his personal record (PR), completing all 26.2 miles in under 4 hours.

Chris' primary inspiration is his son, Will, who was born 13 years ago and diagnosed with HCU at birth. Will has been under the treatment of the amazing staff from Children's Hospital Colorado and Children's Hospital of Philadelphia ever since. Will is doing well, but when a child in the HCU community passed away unexpectedly earlier this year, it made a huge impact on Chris. It was a stark reminder that there is still much work to be done to improve treatment options and ultimately find a cure for HCU.

What do you eat while training for a marathon and how is that different from what you son, Will, can eat?

Chris is logging up to 55 miles a week in his training. As the weekly mileage has increased, Chris is consuming lots of carbs like bagels which are too high in protein for Will's diet. He uses energy gels on his longer runs. Once consumed, these gels are quickly absorbed into the blood to supply calories and nutrients to fuel his runs, delay muscular fatigue, and enhance performance. Most energy gels have zero protein (check the labels) and could be consumed by HCU patients like Will; however, Chris says his son doesn't like them. He also consumes Gatorade to replenish water and electrolytes that are lost during his runs. Gatorade has zero protein and is something that both Chris and Will can agree on.

How can you support Chris' run?

Supporters can go to www.crowdrise.com/o/en/campaign/running-for-rare-boston-marathon-2019/chrishummel1 to make a donation. All the funds raised will benefit HCU Network America's research fund. We also encourage you to make posters/signs in support of Chris' run and post pictures of them to our Facebook page. When adding your posters and signs to social media, use the hashtags #RunforRare, #HCUHeroes #HCUNetworkAmerica #GoChrisGo. Let's show Chris how much his efforts mean to the HCU community!

What are your hopes for the race and for the future?

Overall, Chris aspires to connect with the team, raise funds for a great cause, and take in all the sights and sounds of what he calls "the Super Bowl of marathons". When it comes to running goals, Chris has a very specific one in mind for the 2019 Boston Marathon. He recently became aware of Will Ferrell's (aka Buddy the Elf's) 2003 Boston Marathon time of 3:56:12. Chris is aiming to beat Buddy's time and set a new PR for himself!

For the future, he ultimately wants a cure for HCU, but understands that can take time. In the more immediate future, he hopes to see better treatments and options for HCU patients. Better tasting formula is at the top of his son's list.

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.

3rd International Homocystinuria Meeting

What a fabulous meeting we had at the Third International Homocystinurias Patient Expert Meeting in Rome. The program was packed with the latest research, clinical care and dietary management for these disorders. We are most grateful to the researchers, clinicians and dieticians that attended and for their willingness to spend time and share their knowledge with patients from around the world. We also thank fellow organisers Dr. Carlo Dionisi-Vici, Prof. Henk Blom, Prof. Viktor Kozich and Prof. Ida Schwartz for working with us to be inclusive of patients and allowing them the opportunity to present their perspectives.

The meeting brought together leaders in the field of homocystinuria research, clinical care, nutrition and advocacy to discuss the latest in treatment, diagnosis and research for patients with one of the homocystinurias. The opportunity to bring together patients, caregivers, healthcare professionals and industry is a unique opportunity. As a patient advocate, being able to meet patients and families face-to-face is a delightful opportunity. It was also encouraging to see the medical professionals in attendance genuinely embrace the patient experience and encourage patients and parents to approach them and to talk openly and ask questions.

With over 140 delegates from around the world, approximately 65 were patient/caregivers (making up 40% of attendees), traveling from Australia, Europe, Middle East, USA and South America to be there. This certainly indicates that patients and families are willing to engage with researchers and clinicians to learn from them and also to share their experiences. Feedback from delegates also illustrated the enthusiasm amongst doctors to hear the patient experience first-hand.

The meeting consisted of two days of presentations. The first day sessions covering the following topics: State of the art lectures, Novel developments in remethylation defects, Newborn screening, New developments and an Interactive panel. Day two sessions included the following topics: Long term outcomes, Recent advances in novel treatments, Living with homocystinuria and Patient organisations. The slides presented at the meeting are being collated and will be available, where consent is provided, on the meeting resources section of the HCU Network Australia website shortly.

We will continue to include patients and caregivers in these medical conferences not only in the audience but also as presenters. We truly feel that by involving and including patients in these types of conferences their voice can be added to the discussion and they can be an active participant in improving the lives of patients and their families. Continuing to provide these opportunities for patients to not only engage with each other in a face-to-face setting but also to be involved in these meetings to strengthen partnerships and collaborations in research and clinical care is of upmost importance for HCU Network Australia.

Thank you to everyone who made the Third International Homocystinurias Patient Expert Meeting possible. To all of our speakers and panellists for sharing their work and experience with us, to fellow patient organisations and consortia who collaborated to make this event a success. Thank you to our sponsors, without whom this event would not have been possible. Finally, thank you to all delegates, families and patients, who made this event such a success.

Tara Morrison
Founding Director, HCU Network Australia

Editor's Note: Attendees from the US included Margie McGlynn, President of HCU Network America, who presented an update on the global research map and global grants process for HCU; Dr. Kim Chapman, Board Member and Medical Advisor to HCU Network America who moderated a panel, Dr. Irini Manoli form NIH who presented on remethylation disorders, and Jonathan and Luciana Deveaux, parents of a patient with cobalamin c disorder from California who are members of our community.

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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/

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Click to donate directly