The HCU *Herald*

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HCU HERO: ESMEE FROM ENGLAND

Esmee had her heel prick test at 5 days old and like most parents we didn't give it a second thought. One week later I got 'the' phone call that we will never forget, and over the next 24 hours, we learned that Esmee was the first baby in the South West of England to be diagnosed with Homocystinuria since screening began.

I would compare my initial reaction to grief. I grieved for the uncomplicated childhood that we all wish for our children, and I grieved for the family life that I thought we had lost in some way. It was really tough, and I took the diagnosis very hard in those first few weeks and months. I was lucky to have an extremely positive and hands on husband who totally took it in stride, as well as a strong family that got us through those first few months with hope and optimism. On top of that, we were fortunate to have a fantastic medical team behind us who patiently answered hundreds of my questions/emails/phone calls.

Flash forward and Esmee is a beautiful, happy, healthy and clever 3-year-old who has stormed passed all of her milestones. We are fortunate that Esmee's levels have been consistently stable (illness aside) and that we haven't had any issue with her accepting her protein supplement - in fact she loves it. She is starting to take a real interest in her diet now and she loves telling people that she can't eat animals! She really takes it all in stride with confidence, and she is the happiest, heathiest little girl.

All those fears that I experienced in the early months haven't materialized, and HCU has very much just slotted into our lives - just like any routine or way of life, it's become our normal. Esmee goes to nursery, has playdates, has sleepovers, and gets to experience all of the joys of childhood like any other child. We have been on holiday lots of times as a family, and last year we did a 3-week road trip though the Rocky Mountains and stayed in 8 different places along the way. I can honestly say that HCU never holds us back, and actually it makes us more determined to give Esmee the best childhood.

I personally found my therapy in Esemee's food, and I spent months planning for Esmee's weaning journey. It really helped me through the process and gave me a positive proactive focus to move

forward with. I started an Instagram page, @what_esmee_eats where I share my ideas, and I try to create recipes that can feed the family with a few amendments to suit everyone. It has been amazing connecting with low protein families around the world.

If I could give one piece of advice to other parents, it would be to enjoy your baby and don't allow the diagnosis to rob you of precious moments. It took me about 9 months to come to terms with everything, and after that it was like a light switch moment where the anxiety and emotions melted away. I have my moments - don't get me wrong; but they are far and few between now. It will get much easier and you will adapt.

Tip **
The best advice I got from another HCU mamma was to introduce protein supplement on the spoon at the start of the weaning process before any other food. Esmee's 'first food' was her supplement which she had alongside her low protein infant formula from her bottle. We made sure that she was getting all she needed from her infant formula (bottle), so that the second stage supplement could be given as tastes on a spoon without us having to worry about measurements. I cannot recommend this approach highly enough!

Rare Bears for Kids with HCU

RARE science
Accelerating Cures for RARE Kids

We are thrilled to announce that the Rare Bears for HCU Campaign in partnership with Rare Science has been re-opened!

To enroll in the RARE Bear Program and to request a RARE Bear, please click the link and complete the form: https://www.rarescience.org/hcu/

- Those who have already received a bear, are not eligible
- Date for gifting will be announced later
- You will **not** receive a confirmation email or be notified when your bear has been shipped

Will you be our next HCU patient Hero?

Tell us:

- How you or your child was diagnosed?
- How has HCU affected you, your family and relationships?
- What are some of your successes with HCU
- What are some of your challenges you have faced?
  - How have you overcome them?
- What words of advice would you give to newly diagnosed families?
- For other patient stories, visit: https://hcunetworkamerica.org/patient-stories
- Email your story to: info@hcunetworkamerica.org

START TO FINISH DELICIOUS!

Click picture or name for recipe

APPETIZER

WATERMELON SALAD

MAKES 1 SERVING
SERVING SIZE: 1 SERVING
PROTEIN PER SERVING: 1.5 G
CALORIES PER SERVING: 116

SOUP

NEW ENGLAND STYLE MUSHROOM CHOWDER

MAKES 8 SERVINGS
SERVING SIZE: 4 OZ.
PROTEIN PER SERVING: 1.8 G
CALORIES PER SERVING:118

ENTREE

CAULIFLOWER RICE STUFFING CUPS

MAKES 15 SERVING
SERVING SIZE: 1 STUFFING CUP
PROTEIN PER SERVING: 0.6 G
CALORIES PER SERVING: 26

DESSERT

PUMPKIN PANNA COTTA

MAKES 4.5 SERVINGS
SERVING SIZE: 1 PANNA COTTA
PROTEIN PER SERVING: 1.4 G
CALORIES PER SERVING: 218

YOUR HEALTH MATTERS

MetabERN Recommendations for All Rare Inherited Metabolic Diseases Patients and Caregivers about Treatment Adherence during the COVID-19 Emergency

The COVID-19 pandemic is testing the resilience of robust health systems around the world. This may be impacting you in many different ways, such as creating additional anxiety or exacerbating other medical or therapy issues related to your Inherited Metabolic Disease (IMD). In this difficult period MetabERN underlines the critical importance of sustaining efforts to prevent, diagnose and treat Inherited Metabolic Diseases (IMDs), assuring the continuation of the quality of care provided to you.

To help the entire metabolic community, we MetabERN has developed some recommendations that aim to support all rare IMD patients and caregivers during the COVID-19 emergency.

To find out more, visit: https://metab.ern-net.eu/covid-19/#1585916493939-54217f52-eed3

November Marks Beginning of Open Enrollment for Health Insurance

Do you find your insurance coverage inadequate for low protein foods, formula, betaine, or supplements? Don't fret—November marks the beginning of open enrollment for new health insurance policies.

Feeling overwhelmed? Not sure what policies cover your doctors and your medications? Don't worry, we can assist you with that!

Raenette Franco of Compassion Works Medical is able to assist you with your needs. Raenette can help you find a policy that works for you, or work with your current policy to help you get low-protein foods, medical formula, betaine and "supplements" covered.

There is no fee to work with Raenette, but we do urge you to contact her immediately if you do need a new policy. Open enrollment for 2021 ends December 15, 2021.

You may contact Raenette:
raenettef@compassionworksmrs.com
(973) 832-4736

You are also invited to this webinar to learn more about navigating health insurance. Register now:
https://zoom.us/webinar/register/4416033065020/WN_MPhF-s6dQwaYVdWjMp5ybA

NORD Rare Disease Parent Respite Program

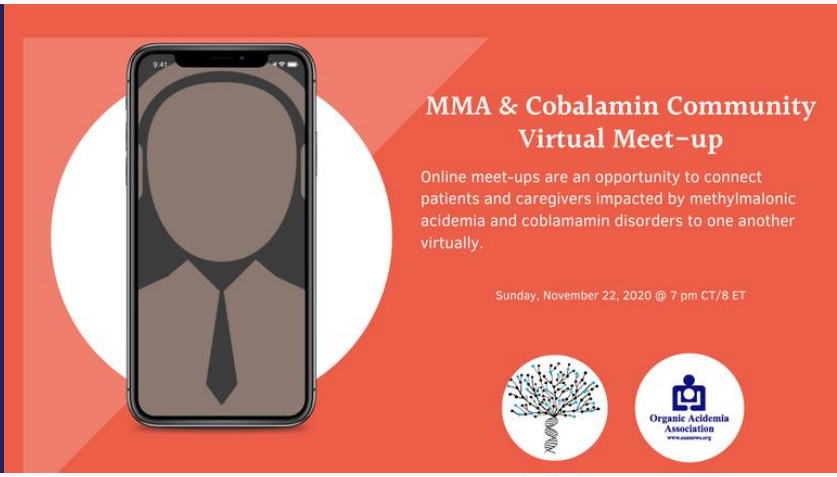
Caring for a loved one with a rare disease demands significant time, attention, patience and dedication. NORD's Rare Caregiver Respite Program is designed to give back to caregivers—the parent, spouse, family member, or significant other—of a child or adult living with a rare disorder, and provides financial assistance to enable the caregiver a well-deserved break.

Through the program, NORD will provide financial assistance to eligible caregivers covering up to $500 annually so that a respite caregiver may be secured to care for a loved one. Potential applicants can reach out to NORD to ascertain if they meet eligibility requirements. The grant may be dispersed throughout a calendar year or be used in a single award. Examples of what the NORD Rare Caregiver Respite Care Program financial assistance may cover include:

- Registered Nurse (RN) care
- Licensed Practical Nurse (LPN) care
- Certified Nursing Assistant (CNA) / Nursing Assistant
- Home Health Aide (HHA)

The fund continues to accept additional donations to ensure this first-of-its-kind program is sustained for years to come. For more information and instructions on how to apply or donate, visit: https://rarediseases.org/wp-content/uploads/2019/05/NRD-1182-RespiteCare_SS_1up_FNL_NoCrops_Fixed.pdf

We All Need Support; Come Check Out Our Virtual Meet-ups!



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

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

INCASE YOU MISSED IT!

Raising Awareness: See How We Engaged the FDA!

On June 26, 2020 a group of four HCU patients and caregivers, along with HCU Network America's President (who lost 2 sisters to HCU), and a metabolic geneticist (lead author on the Guidelines for Diagnosis and Management of Cystathionine Beta-Synthase Deficiency) held a "Patient Listening Session" with the Food and Drug Administration (FDA) to help the regulators understand the issues and unmet needs of the patient community. The group provided personal and emotional first-hand testimony of what it is like living with and caring for someone with this rare and sometimes fatal disease.

The main topics discussed during the listening session were:

- Communicate struggles patients face with current treatments approaches, especially dietary restrictions
- Share how HCU directly affects our health and daily lives
- Convey how important a new therapy is that can reduce dietary restrictions and prevent clinical issues

Read our executive summary for the more information: https://2d3.505.myftpupload.com/wp-content/uploads/2020/09/FDA-Listening-Session-Summary-Final.pdf

Retrophin Announces Agreement to Acquire Orphan Technologies
October 22, 2020 - Orphan Technologies announced that it was being acquired by Retrophin, Inc. Retrophin has 3 products on the market today and is committed to bringing new therapies to the rare diseases and ultra-rare disease community. With this acquisition, the ongoing and planned studies and timeline for OT-58 remains the same. Retrophin will continue with their current phase 1/2 trial and opening of new sites once clearance is allowed given the current COVID impact. This should be a seamless transition between companies. Retrophin takes a proactive approach to collaborating with the patient and medical communities so you should expect a high level of engagement and listening to continue. Importantly, similar to Orphan Technologies, the entire organization at Retrophin will be fully committed to advancing OT-58 as quickly as possible for people living with HCU. HCU Network America looks forward to the collaboration between Retrophin, Inc and welcome any questions the community may have about this.

Read the full press release here

Aeglea BioTherapeutics Announces Orphan Drug Designations for ACN00177 for the Treatment of HCU
October 26, 2020 Aeglea BioTherapeutics announced that the FDA and the European Medicines Agency have granted Orphan Designation to their investigational therapy for Homocystinuria for the US and EU, respectively. Learn more here: https://t.co/DBJq5OxxvE

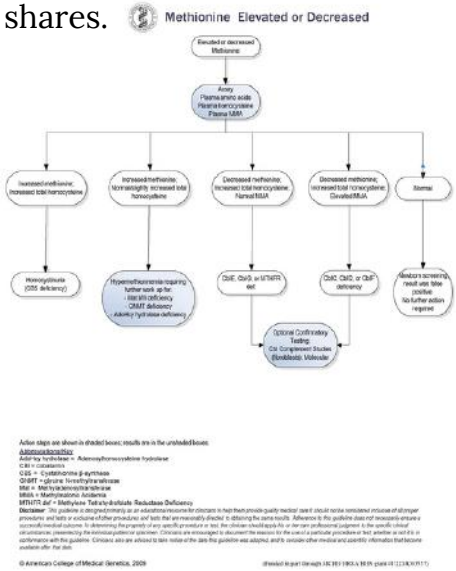
HCU Awareness Month Recap

Social Media Reach

Social Media is a powerful tool when you are using it to raise awareness. Not only does HCU Awareness Month bring attention to our organization and the actual disease, but it also draws attention to other issues related to homocystinuria. We see a lot of growth thanks to those who follow along with us and share. During the month of October we gained 39 Facebook followers and 7 Twitter followers. Over the course of the month, we reached thousands of people! We can't thank our community enough for participating in HCU Awareness month!

Typically we look at post with the most views, but this year we thought we should highlight post with the most shares, so we are highlighting the fact post and the activity post with the most shares. With 15 shares, our HCU Awareness 2020 Day 1 fact had the most shares.

#HCUAwareness2020: Fact 1: #Homocystinuria is an elevation of the amino acid, #homocysteine (one of the building blocks of protein) in our urine or blood. High homocysteine can be caused by #Cystathionine-Beta-Synthase (relates to B6), #Cobalamin disorders (relates to B12), remethylation disorders, such as #MTHFR (relates to Folate ((which is B9) and B12).



With 14 shares, our HCU Awareness 2020 Day 27 activity post had the most shares.

Day 27 of #HCUAwareness2020 : All states Newborn Screen includes Classical Homocystinuria and most states screen for Coabalmin C. Low methionine also may trigger investigation and diagnosis of the other more rare Cobalamin disorders and Severe MTHFR

Until ALL homocystinuria patients are screened for accurately at birth, we will continue to speak up for better newborn screening (NBS) in homocystinuria patients.

What can you do? All NBS programs are at risk - contact your legislators and ask them to co-sponsor H.R. 2507/ S. 2158, the Newborn Screening Saves Lives Reauthorization Act! Every day this isn't passed, Newborns lives are at risk! https://everylifefoundation.org/voter-voice/

Thank you to everyone who participated in activities and shared during our 2020 HCU Awareness Month!

Fabulous HCU Awareness Fundraisers!

Top row, left to right: The Sullivan Family, The Carter Family, The Smeltser Family
Middle Row, left to right: The Mayes Family, The Koelling-Franqui Family, Kelly Waters
Bottom Row, left to right: Joann Ball, Samantha Mozden, Pamela Penrose

Thank you to the 9 families who fundraised on behalf of HCU Awareness Month!
From Facebook donations alone, we have raised $4,456!

What is Amazon Smile?
Amazon Smile is a simple and automatic way for you to support HCU Network America every time you shop, at no cost to you. When you shop, you'll find the exact same low prices, vast selection and convenient shopping experience all with the added bonus that Amazon will donate a portion of the purchase price to us.

How do I set it up?
Simply, go to smile.amazon.com, the first time you enter the site it will ask you to designate an organization. Type in HCU Network America and select us from the list. It is important to note that in order for the donations to go to HCU Network America, you MUST check out from this url every time - see best practices below for some pointers on how to do this.

What if I'm already set up and would like to switch to HCU Network America?

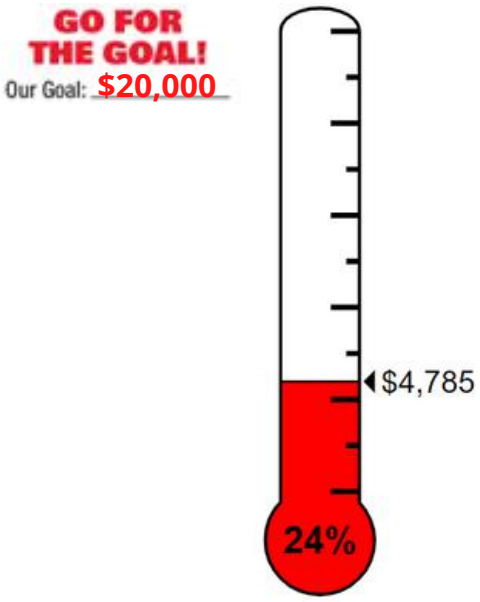
1. From your desktop, simply select "Your Account" from the navigation at the top of any page
2. Then select the option to "Change your Charity". From your mobile browser, select "Change your Charity" from the options at the bottom of the page.
3. Type HCU Network America in the search bar and search for the charity.
4. Select HCU Network America charity to update your account

If you are still having trouble, visit https://hcunetworkamerica.org/amazon-smile/ for the steps with images of how to.

Best practices for using Amazon Smile on a desktop
Now that your account is set up to use Amazon Smile, it is important to note that Amazon only makes donations to HCU Network America when you checkout from your cart from this smile.amazon.com. This is the only way HCU Network America gets any donations from Amazon Smile.

Shopping from your phone? Android and iPhone users, rejoice – you can now shop Smile.Amazon from the app – check out the instructions here- https://www.amazon.com/b?ie=UTF8&node=15576745011

$20,000 DONOR MATCH IS BACK!



That's right, you heard us right! Thanks to an anonymous donor, any **funds** you help raise from October through December 31, 2019 **will be matched up to $20,000!**

Don't forget to submit your donation receipt to your employer so it can be matched!

We are asking every patient and family to help us raise funds for homocystinuria. During the winter holiday's warm hearts and generosity can be felt near and wide. During this time, we ask that you share our appeal letter with your colleagues, friends and family.

See our appeal on the next two pages!
You can also print it here

Here are the steps to see if your company matches gifts!

Keep an eye on your inbox; we will be emailing details out shortly!

HCU Network America
15 S Mallory Ave
Batavia, Illinois 60510

(630) 360-2087
info@hcunetworkamerica.org
http://www.hcunetworkamerica.org
Tax ID Number: 81-3646006

The year 2020 has been an extraordinary challenge to us all. Everyone at HCU Network America hopes that you and your loved ones are managing to get through this safely, and if you have suffered from COVID-19 illness or losses in any way, you have our deepest sympathies.

These times are extra demanding for people with unique medical needs and special diets, who spend time at clinics and waiting for life giving medical supplies. People with HCU have to balance all the variables in their lives that affect their health and that is tough, especially this year, and can sometimes seem overwhelming.

HCU Network America is here to help this special group of people with the support and resources they need to navigate daily life. We are proud of the reach we have and the way these communities have knit themselves together. But we can't do it without your help! As a 501(C) (3), we need your donations, which are tax deductible, to continue with our mission and meet our goals. What you do to help us, can make a huge difference in the lives of all HCU patients and their families.

We would like to share a story of a family's journey of diagnosis.

> *Our daughter had just turned 3 in the spring of 2012 when she suffered a large blood clot on the brain and a series of small strokes. After a week's stay in a children's hospital we were released without any explanation as to the cause of the large blood clot. Six weeks later, during our follow up, the doctors ordered extended blood tests which resulted in a test coming back with elevated homocysteine levels. The level of homocysteine in our daughter's blood was so high that it was actually toxic to her body and prone to clot. It was shortly after this test result came back that we had our explanation to the blood clot as well as a new lifelong diagnosis - our daughter had Homocystinuria. During our crash course on this rare metabolic disorder we learned that she was born with this & that it is hereditary - it was recommended that we have our 18 month old son tested as well. His results came back positive - both of our children were diagnosed with HCU. Homocystinuria was missed on initial newborn screening for both of our children and the state we reside in doesn't offer a second newborn screening where HCU should be flagged. Thankfully the doctors were able to diagnose our daughter correctly after her event and fortunately we were able to get our son on the treatment regimen preventing any events for him. So began our journey with Homocystinuria.*

You can imagine what that felt like and at that time there was no HCU Network America to give this family support and vital information. In June of 2016, HCU Network America was incorporated; bringing hope to families living with HCU that they had advocates to help them get the latest and best advice from the medical community. HCU Network America also financially supports research that can help find new treatments. Since 2016 we have communicated with metabolic clinics all over the country to reach out to new patients and provide them toolkits which are filled with helpful tips and guidelines for living with HCU.

Here are some of the highlights that your donations helped with:

1. Help fund Research Grant: In 2020, HCU Network America, in partnership with HCU Network Australia, issued its second research grant in the amount of $40,000 to Dr. Tomas Majtan, PhD from the University of Colorado Anschutz Medical Campus, Aurora for the evaluation of benefits of thiol-based reductants in classical Homocystinuria
2. Expand understanding and awareness of the need for new therapies for Classical Homocystinuria by conducting Patient-Listening Session with the FDA
3. Distributed Improved Newborn Screening Talking Points for the Homocystinurias to medical professionals and professional Inherited Metabolic Disorder Organizations
4. Helped secure session on improved Newborn Screening for the Homocystinurias at the Association of Public Health Laboratories 2020 Newborn Screening Symposium
5. Published an Emergency Planning Guide with Emergency Protocol Template for all Homocystinurias
6. Published educators guide for Cobalamin Disorders and MTHFR
7. Provide a consultant with experience in the insurance industry to fight for coverage for medications and food at no cost to HCU patients
8. Support for bi-monthly community meetups

Our goal this year is $50,000. Thanks to an anonymous donor, any funds you donate up to December 31, 2020 will be matched up to $20,000. Please consider a donation to HCU Network America in 2020. We need your help and appreciate any donation. In addition, if your employer matches charitable donations, they will match those too!

Take a minute to look at our website to see what we are up to and meet some our "heroes": https://hcunetworkamerica.org. You can donate through our website or by mail.

Thank you for all you do to help us - we will all get through this year and we look forward to 2021!

Thank you,
Danae' Bartke
HCU Network America, Executive Director

Donor levels:

- Leadership Circle - $5,000 or more
  Donor's name, HCU Patient's name and photo on homepage of website, along with certificate donation

- HCU Champion - $1,000 or more
  Donor's name, HCU Patient's name and photo on HCUNA donation page along with donation certificate

- HCU Supporter - $500 or more
  Donor's name, HCU patient's name on website, along with a donation certificate

- HCU Ally's - $100 or more
  Donors name on website along with a donation certificate

- HCU Friend - $99 or less
  Donor's name listed on website

Live better, together!

Join our Contact Register and Patient Directory!

What is the contact register?
The contact register is a secured private survey that allows you to share information on you or your family member with HCU with us. This includes where you are from, your relationship to homocystinuria, the patient's birthdate, gender, their exact diagnosis (e.g. CBS, cobalamin, or MTHFR), how they were diagnosed, and if the patient was diagnosed through newborn screening. This information is kept confidential and will not be shared unless you give us permission. By registering, you will be able to identify other patients in your state and request their contact information. You will also be able to access information posted over time that can only be shared with the patient community. (For example, we may have webinars that the expert presenter does not want to be publicly available, but is willing to share with the HCU community.)

What will this information be used for?
HCU Network America strives to inform patients and families with resources, create connections, and support advancement of diagnosis and treatment of HCU and related disorders. The information you provide helps us succeed in our mission - plan events, develop resources and educational tools, and ensure everything is being done to support timely and accurate diagnosis from birth. It also allows us to have informed conversations with doctors, pharmaceutical companies, and law makers. Your information helps us understand the landscape better so we can better advocate for you!

How do I participate?
The contact register form takes approximately 3-5 minutes to complete. You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to: https://hcunetworkamerica.org/contact-register/

Trying to figure out a perfect gift for someone cooking with the low protein diet in mind? Check out this great list of ideas for every level cook! https://pkunews.org/kitchenwishlist/