The HCU Herald

Featuring...

HCU Hero

Sienna from the UK

HCU AWARENESS MONTH EDITION!

HCU NETWORK AMERICA

All things Homocystinuria: patient stories, resources, research, events and more!

HCU HERO: SIENNA FROM THE UK

Sienna's Story

On the 16th September 2009 we were in the hospital with my son who was suffering from breathing issues. At 16.30 my phone rang and it was my mum. “Stu, you need to speak to the optician”. My mum had taken our then 5-year-old daughter Sienna to Specsavers for a routine eye exam. She had been squinting a lot and sitting right in front of the screen to watch TV so we thought she might need glasses. The Optician said that Sienna had dislocated lenses in her eyes and it was at that moment our lives changed forever. Left alone with “Google”, we went down a rabbit hole of what dislocated lenses could mean. We realised that it meant an operation, so we researched the leading eye surgeons in England where we found a specialist in Bristol. The next week we made an appointment to go and see her for a consultation and she said that given that dislocated lenses usually meant Marfan Syndrome, Sienna would need to have a heart check up to make sure her heart was ok to undergo the anaesthetic - yet more worry!!! When we saw the heart specialist the following week, he said that her heart was fine, but wanted to do another test as at the very back of his mind, he had a nagging suspicion that he wanted to rule out. We later learned that the "nagging suspicion" was Homocystinuria.

He had a nagging suspicion that he wanted to rule out. We later learned that the "nagging suspicion" was Homocystinuria.

The following week we had a call to confirm that the sample he had taken suggested that it was indeed HCU and that Sienna would need another blood test (the first of lots over the coming years!!) carried out to confirm the potential diagnosis. We would also have to have her younger brother tested to see if he had it too, which of course caused even more worry.

Then in December I had a call from Dr. Germaine Pierre at Bristol Childrens Hospital to confirm that Sienna's blood test had confirmed the diagosis of HCU, whilst our son's test showed a negative result. Our local specialist hospital, Bristol, was a 400 mile round trip for us from home, and I've lost track of how many visits we had to make there and back over the years. Whilst we waited for Dr. Pierre's confirmation, Sienna had to have her eye operations. Once Sienna was under her first anaesthetic, I remember feeling completely helpless and all the bottled up emotions of the past few weeks came out and I recall just asking why this was happening to our little girl.

Thankfully the operations were a success and I remember walking with Sienna whilst she was holding her mum's hand and staring at the back of it. I asked her what she was looking at and she said "the dots". I gathered she meant it was the freckles on the skin that she could see and then realised that she had never seen life in detail before. Another really emotional moment!

As part of the lens removal operation, Sienna had contact lenses put in and whilst it was great that she could see clearly, they definitely caused some issues! Whilst in hospital having a lens replaced, Sienna wouldn't allow them to do it and some assistants forcibly held her so it could be done.

I remember her screaming as they tried for what was only a few seconds but felt like forever to remove it. I was just about to tell them to stop when they got it out, but she now still can't allow anyone near her eyes and has been diagnosed with PTSD as a result. It's a similar thing with needles from all the blood tests she has had and remained a major issue recently with her having her Covid Vaccines - taking about 30 minutes on each occasion before she would let them inject her.

As a parent, the most difficult thing was just how helpless we were with it all. Our little girl had to have all of these surgeries, tests, exams and there was nothing we could do. It didn't help that there was no support or other people that we could really speak to and we felt cut off, isolated and alone. I think however that this gave us some real internal strength and I found over the coming years that we had to be a very strong (and loud!!) voice in advocacy for Sienna both medically and educationally.

I found over the coming years that we had to be a very strong (and loud!!) voice in advocacy for Sienna both medically and educationally.

We have certainly had a few arguments in getting the best we can for her and I recall telling a doctor that Sienna may be their patient but she is my daughter and I will always advocate for her and if that was a problem, then tough!

In a period of reflection I remember reframing my view on the diagnosis. Obviously we would not want Sienna to be diagnosed with HCU, but having read of the life expectancy undiagnosed, we soon realized that the diagnosis, treatment and medication ultimately saved her life. I wrote a note to that heart specialist who had had the "nagging suspicion" and told him that he had saved my daughter's life and no words could express my thanks.

There have also been some amazing experiences that have happened as a result. We've met some great people from all over the world, experienced fantastic support from those with PKU on dietary advice and met and received care from extraordinarily dedicated professionals. Sienna was granted a "wish" by the Make-A-Wish foundation and she wanted to go to Disneyland and meet the Princesses. When I told her how Make-A-Wish works, Sienna wanted to raise some money as a thank you donation and set up a virtual lemonade stand. This got picked up by the local paper and eventually she ended up raising £1296.

As part of the paper's story, we said how she wanted a Disney Princess dress but was too large to wear the children's dresses that you could buy. It's funny how the universe works as we were then contacted by a seamstress who offered to make Sienna a custom dress to wear to Disneyland - an amazing gesture and Cinderella did indeed go to the ball!

13 years ago, the diagnosis presented a very limited future, but over these years, the progress in medicine (and definitely in low protein food!) has improved massively. I'm so glad that they were able to find a replacement for the HCU coolers - those sickly orange drinks. I remember tasting them at the time she first got them and I think I can still taste that horribleness today! I don't know how she managed them for all those years.

I am so proud of Sienna and how she has, and continues to, just get on with it. She's always smiling despite all the difficulties and always looks at what she can do instead of what she can't. I am not sure if it's irony, coincidence or a bit of both, but on 16th September 2022 - 13 years to the exact day of that fateful optician's trip - we dropped Sienna off at University to start the next chapter in her life - studying for a degree in Musical Theatre.

Who knows what the future has in store but if I think back to 2009, I would never have thought that in 13 years she would be off to University, but here we are. I can only imagine what fantastic things she will do in the next 13 years - I just hope they don't go so fast as the last 13!

Whatever she does, she will always be my HCU hero and inspiration.

This HCU Hero story was written with love by Stuart, dad of Sienna.

HCU HERO: SIENNA FROM THE UK

Meet Sienna!

- I have just arrived at University where I am going to be studying for a degree in Musical Theatre and I'm really proud that I have managed to get here.
- My passions include singing, drawing, watching anime and performing. I'm also really passionate about Japan and I hope to be able to go there one day.
- I am hoping that sometime soon I will be able to eat what I want and not be limited to 10 grams of protein a day. And, no more needles!

Growing up I was not allowed to fly because I was told that it would put me at an increased risk for blood clots. So, my parents would book cruises instead. These were fantastic because on board I would get my own chef! I would take all my low pro mixes with me and then every night I would order food from my recipe books and they would make it for me and it was always fantastic. Sometimes they wouldn't realise that it was just me and would make a whole cake or a dozen ginger bread men just for me.

Within the last few years I was told that I could go on a short haul flight, and as the annual HCU Network Australia conference was in Rome 2019, this meant that I would be able to attend! I was really excited about flying for the first time and Dad said that if I was going to fly, then it would be best to fly to a place where all the best HCU doctors in the world would be. This was also the first time that I was actually able to meet someone else with the HCU and for once I didn't feel like I was the only person in the world with HCU.

WEAR YOUR AWARENESS

October is HCU Awareness Month!
Get your Gear NOW!

https://www.bonfire.com/store/hcu-haberdashery/

All proceeds from the sale of our gear benefit HCU Network America!

GO BLUE FOR HCU

October is HCU Awareness Month

HCU Network America — Hcunetworkamerica.org — info@hcunetworkamerica.org

It's HCU Awareness Month –
LET'S TALK FUNDRAISER SETUP!

There are many ways that you can participate in HCU Awareness Month.

One way is by hosting a fundraiser!

Not only do fundraisers raise vital funds to help us support the Homocystinuria community, they spread awareness of the Homocystinurias and the challenges of living with the conditions.

Not sure how to get started?
Let our Fundraising committee help!

Email info@HCUnetworkamerica.org

Click here for a list of additional HCU Awareness and Fundraising Event Ideas!

HCU Awareness Month

Activity List

We know this is a rather long list, but we love your participation as it helps raise awareness for our small but mighty community. Please try to do as many as you can. If you do one each day, you'd complete the list!

- ☐ Change your social media picture to the HCU Awareness Ribbon
- ☐ Start a HCU fundraiser
- ☐ Share an infographic about HCU
- ☐ Share a patient story
- ☐ Share your diagnosis story
- ☐ Challenge your friends to the same amount of protein and three normal protein shakes a day #ToastTocHCU
- ☐ Share a pic of an item that has the same amount of protein you can have
- ☐ Share your daily diet record –completed
- ☐ Share a low-protein meme
- ☐ Share your favorite low protein recipe! Bonus if you cook it and share a pic
- ☐ Dining out, low protein style. Where do you like to eat?
- ☐ Share a pic of what your grocery store haul looks like
- ☐ Real cost of HCU: Grocery Cost Comparison #Medical Nutrition Equity Act, or share some patients with HCU require injectable B12. B12 on average is $300-400 a month and most insurance companies don't cover it!
- ☐ Share a picture or video capturing all the medication you take (this includes formula for those who need it).
- ☐ Share a picture of your first pair of glasses, or a device that helps you navigate or communicate due to lack of vision
- ☐ Share something you wish people understood about HCU
- ☐ #HaikuforHCU—Write and share a Haiku describing life with HCU
- ☐ Wear jeans for your rare genes #ItsInOurGenes
- ☐ Wear your HCU Shirt and share a pic online—#HopeConnectsUs
- ☐ #GoBlueforHCU
- ☐ #HCUAwareness post in a public place
- ☐ Share with a stranger what HCU is and why it's important to you
- ☐ #Create4Cure—Create a work of art that brings awareness for HCU— can be a song, dance, a painting—get creative!
- ☐ #High5forHCU—List 5 ways HCU makes you a stronger, better person
- ☐ All states test for classical HCU, but many are still missed
- ☐ Share a picture of you and a HCU buddy! Or tag a friend who is of great support
- ☐ #FacesofHCU—Share a picture of you saying, I am one of the 1 in 200,000 people with HCU
- ☐ #Hope4HCU—Share 4 things that give you hope and encouragement
- ☐ Share the HCU timeline—if you know other facts, let us know!
- ☐ Cutting Edge of HCU: Share about a therapy that is in the works!

To find additional information and resources, visit:
https://hcunetworkamerica.org/hcu-awareness-month/

OUR MATCHING GIFT IS BACK - BIGGER AND BETTER THAN BEFORE!!!

That's right, you heard us right! Another anonymous donor has joined (we went from 2 to 3!). That means that **any funds** you help raise from October through December 31, 2022, **will be matched... up to $30,000!**

We are asking every patient and family to help us raise funds for homocystinuria. Set up a Facebook or Instagram Fundraiser, Give Lively, GoFundMe, or host your own alternative fundraising event and invite your family and friends to participate! Alternatively, they can donate directly to HCU Network America. **Anyone who creates a fundraiser and raises over $100, will receive a HCU Awareness car magnet!**

Have an idea for a fundraiser, but not sure how to get started? Let our fundraising committee help you get started Email info@hcunetworkamerica.org and we will connect you!

EMPLOYER MATCHING GIFT PROGRAM

Did you submit for your Employers Corporate Matching Gifts Program?

What are Matching Gifts?
Corporate matching gifts are a type of gift giving in which companies financially match donations that their employees make to nonprofit organizations. When an employee makes a donation, they need to request the matching gift from the employer, who then makes their own donation. Companies usually match at a 1:1 ratio, but some will match 1:2 or 2:1.

Why are Corporate Matching Gifts Valuable?
Corporate matching gifts are valuable because they are free money for your nonprofit of choice, HCU Network America! Your donation has double the power without you having to give double the amount.

Does HCU Network America Really Benefit?
If corporate matching gifts weren't valuable, we wouldn't be sending this letter. In 2020 we received approximately $45,000 in corporate matching gifts!

How Do I Find out if my Employer has a Corporate Matching Gifts Program?
Your company website or websites such as doublethedonation.com make it easy for you to search and see if your employer takes part in corporate matching gifts. If you can't find it online, please consult with your employee handbook, HR rep or manager to find out.

How Do I Request my Donation is Matched by my Employer?

1. The donor completes their donation
2. The donor submits matching gift request
3. Company reviews donation and nonprofit eligibility and reaches out to nonprofit
4. Nonprofit verifies the donation was made
5. If eligible, the nonprofit will receive the matching gifts request!

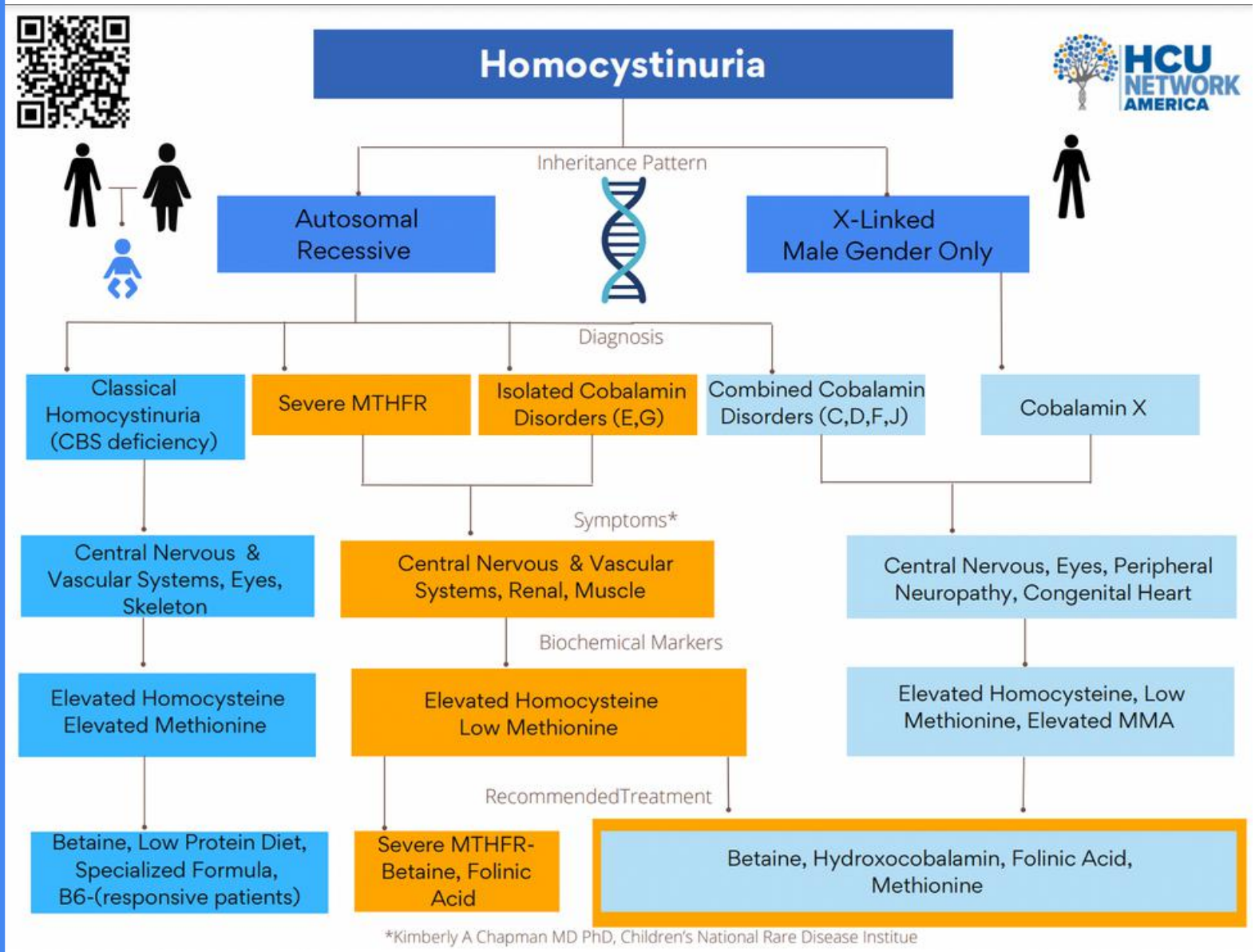
Top Matching Gift Companies

Company	Match Ratio
General Electric	1:1
Gap Corporation	1:1
ExxonMobil	3:1
Johnson & Johnson	2:1
Microsoft	1:1
Pfizer	1:1
Coca-Cola	2:1
And many more!	

Did you know some companies match retired employees donations?

NEW RESOURCE

HOMOCYSTINURIAS FLOW CHART



Download here: https://bit.ly/HCUFlowChart

NEW RESOURCE

MTHFR

Could it be Homocystinuria?

Common Variants	Rare Variants
• MTHFR deficiency (C667T and A1298C) • Found in 30-40% of the U.S. population	• Homocystinuria due to severe MTHFR deficiency • Fewer than 100 documented cases worldwide
• MTHFR enzyme activity only mildly or moderately reduced	• <1% MTHFR enzyme activity
• Homocysteine - normal or slightly elevated; normal range is 5-15 (µmol/L) • Methionine - normal	• Homocysteine - higher than normal; often over 100 (µmol/L) in untreated individuals • Methionine - usually lower than normal
• No treatment typically recommended	• Treatment must be prescribed by a doctor
• **Symptoms** Typically not symptomatic	• **Symptoms** Early-Onset: Lethargy, Feeding Problems, Seizures, Global Delays Late-Onset - Cognitive Deficits, Behavior Problems, Seizures

Download here: https://bit.ly/HCUMTHFR

PUMPKIN APPLE CRISP

Serving Size: 125 g | Protein per serving: 0.7 g | Calories per serving: 244

Ingredients:

- 4 c Chopped Apples 32mg
- 1/4 c Brown Sugar 2mg
- 1 tsp Cinnamon 3mg
- 1/4 tsp Ground nutmeg 1mg
- 1/2 tsp Ginger, ground 2mg
- 1/2 tsp Salt
- 1 TBSP Cornstarch 1mg
- 1/2 c Canned Pumpkin 44mg
- 1 tsp Vanilla Extract

Crumb topping:

- 1 c Cook for Love Baking Mix 76mg
- 1/2 c Brown Sugar 4mg
- 1 tsp Cinnamon 3mg
- 1/4 tsp Salt
- 5 TBSP Butter, melted 30mg

Directions:

1. Preheat oven to 350 degrees. Spray a casserole dish with cooking spray.
2. In a large bowl add the apples, spices, cornstarch, vanilla, and pumpkin. Mix to combine and everything is well coated. Pour into prepared casserole dish.
3. Prepare crumb topping by combining the CFL baking mix, brown sugar, cinnamon, and salt. Lightly whisk to combine. Add the melted butter and use a fork to combine. This will help make the crumbs. Pour crumb mixture on top of the apple filling. Bake for 45 minutes. Use a paring knife to test the tenderness of the apples. If the knife comes out easily, remove the crisp from the oven. Allow to cool 10 minutes before serving.

THIS WEEK'S MENU

Each day has meals for <10 grams (g) of protein/day, 20-30 g. of protein/day, and 30-40 g. of protein/day.

M

Breakfast: Breakfast Sandwich
Lunch: Portobello Mushroom Wrap
Dinner: Mac & Cheese & Steamed Vegetable Medley

T

Breakfast: Fruit Breakfast Bowl
Lunch: Tacos
Dinner: "Burger"

W

Breakfast: Banana Muffin and Yogurt
Lunch: Veggie Wraps
Dinner: Spagehtti & Veggie Meatballs

T

Breakfast: Waffle & Fruit
Lunch: Veggie Nuggets
Dinner: "Ricotta" Lasagna & Texas Toast

F

Breakfast: Country Breakfast
Lunch: Grilled Cheese Sandwich & Creamy Tomato Soup
Dinner: Pasta Alfredo & Broccoli

Shopping List

Click each day to view the week long menu!

Disclaimer: This meal plan is intended to be a foundation or guide to what meals could look like on a low protein diet. It does not take into account individual caloric, protein and formula requirements, which are all patient-specific. Please consult with your metabolic geneticist and dietitian prior to making any significant dietary changes or following any meal plans of which you are unsure.

Vitaflo® Recipe

Spooky Skull Cookies

Preparation time: 10 minutes
Baking time: 20–25 minutes
Recipe makes: 8 cookies

Ingredients

3/4 cup margarine, softened at room temperature

1/4 cup sugar

1 1/2 cups (165 g) low protein all-purpose mix

1/3 cup corn starch

1/2 tsp vanilla extract

Permitted decorations: such as fondant, edible icing pens, colored sugar, and sprinkles*

* Decorations are not included in the nutrition information.

Directions

1. Preheat oven to 320°F.
2. Combine all ingredients in a large bowl, mixing well until a dough forms.
3. Cover a baking tray with parchment paper.
4. Using a rolling pin, roll out the dough between two sheets of parchment paper.
5. Cut out 8 skulls from the dough using a skull-shaped cookie cutter and place on the baking tray.
6. Bake in preheated oven for 20 minutes or until cookies are golden brown.
7. Remove cookies from baking tray and place on a wire rack to cool.
8. When completely cooled, decorate with permitted decorations.

Nutrition Info Per Serving (1/8 recipe):

Calories: 270 | Protein: 0.1 g | Phenylalanine: 5 mg

Please check the suitability of this recipe for you with your healthcare professional.

This recipe was analyzed using HowMuchPhe.org.

For more recipes, please go to our website **www.VitafloUSA.com** and follow us on Instagram ***@vitaflousa*** and on facebook ***@Vitaflo Vitafriends.***

This recipe has been specifically designed for a low protein diet. The nutrition information for a recipe could change depending on the brand of product you choose and should only serve as a guideline. Refer to labels for allergen information and suitability.

VFUSA_SPOOKYSKULLCOOKIES_102020

Vitaflo®

Innovation in Nutrition

A Nestlé Health Science Company

Fall foods are ready in no time and will warm you up


Pasta Duets™ Mac & Cheese


Toasted Pierogis


Gingerbread

Visit **cambrooke.com** to find a variety of easy, tasty foods and hearty recipes that will keep you warm this season.

Try Homactin AA Plus for HCU

- **Great Refreshing Flavor:** Lemon Lime
- **Flexible for all ages:** 15g PE & 150 kcals
- **Low Volume:** Mix with 5 oz. water
- **Optimized Bone Health Profile:** Vitamin K2 and - PRAL

Cambrooke is excited to support the HCU Community

To request a sample visit **samples.cambrooke.com**
To place an order call **Customer Service** at **866 456 9776, option 2**

Eat Well, Live Well.
AJINOMOTO

CAMBROOKE™

RECORDATI CLICK CAMPAIGN RECAP

872 Clicks 4 HCU

Thank you **for clicking!**

And a very special "thank you" to
Recordati Rare Diseases
for sponsoring our "Click Campaign"!

RECORDATI RARE DISEASES GROUP
Focused on the Few™

Thank you!

to our Race for Research sponsors!

aeglea™

CODEXIS®

RECORDATI RARE DISEASES GROUP

Focused on the Few™

Meet our Fabulous Fundraisers!

Brooke's Blazers Severe MTHFR fund	$3,150
Cakes for Carson - Severe MTHFR research fund	$4,010
Carter Crew 4 HCU!	$175
Ellie's Entourage	$3,575
Grayson's Gang - CblG Research Fund	$2,156
Joanna Ball	$250
Masen's Movement!	$2,311
Mighty Marchese's	$1,100
Miles for Marley - CblC research fund	$760
Mississippi Madness	$250
Run with Shu	$200
Sylvia's Supporters - Recordati	$360
Synlogic	$70
Team Aeglea	$325
Team CblG - Carson Hunt	$16,385
Team Codexis	$920
Team Hawkins	$1,900
Team Recordati	$331
Team Will for HCU	$4,433
The Bartke Ruff Ruffs	$575

A huge THANK YOU to race participants & fundraisers!

FEATURED FUNDRAISER

Team CblG - Carson Hunt

Carson's fundraiser raised an astounding $16,385 for Cobalamin G research!

CblG is a rare form of Homocystinuria. With only 50 identified cases worldwide in the literature, the need for research is critical.

A very special THANK YOU to **Sigma Phi Epsilon** Fraternity at the University of Arkansas!

Lead by Ethan Hunt, Carson's brother and SigEp member, the fraternity hosted their own successful fundraising efforts for Team CblG!

Want to feel inspired? Check out Carson's story on our Youtube channel!

https://bit.ly/3fmCXzR

HCU IN THE NEWS!

Riding for a cause

BVNW student takes fundraising into his own hands, raising money towards research for a rare genetic disorder.

Reagan Kauth, Writer | September 15, 2022

At 3 months old, freshman Carson Hunt was diagnosed with Homocystinuria (HCU) Cobalamin G (CBLG), a rare genetic disorder. This disorder has caused Hunt's levels of the amino acid homocysteine to increase far beyond a typical amount, which has led to blindness, developmental delays and may pose other potential health issues in his future.

Freshman Carson Hunt began riding his bike to help raise money to fund Homocystinuria (HCU) research. (Lila Vancrum)

Freshman Carson Hunt poses alongside his parents, Dana and Darren Hunt. (Lila Vancrum)

In order to help other families dealing with HCU, Dana helped create the HCU Race for Research in 2019 to raise funds for researching the rare condition. The Race for Research will take place throughout the entire month of September. During this month, Hunt will be riding his bike 100 miles to help raise money to donate.

Read the full/original publication here! bit.ly/3LjW3m8

SPECIAL EDITION

FASEB hosts *Folate,Vitamin B12 and One Carbon Metabolism conference* in Asheville, NC August 14–19, 2022.

Brittany Parke, an HCUNA board member and cobalamin steering committee lead, recently had the opportunity to participate in a round table discussion at the FASEB Folate,Vitamin B12 and One Carbon Metabolism conference in Asheville, NC. The discussion centered around the management of inherited B12 disorders which included inborn errors of cobalamin metabolism (cblA, cblB, cblC, cblD, cblE, cblF, cblG, cblJ, cblK and clbX). Along with Brittany, Dr. Charles Venditti was part of the round table discussion and explained the complexity and difficulty of diagnosing and managing disorders of cobalamin metabolism. Participants were interested in the high doses of Hydroxocobalamin given to patients and were surprised to hear the difficulty that patients encounter in getting the medication from compounding pharmacies paid for by insurance. Brittany was able to present on the patients perspective and the many areas of unmet need that were highlighted in the recent completion of the research map and strategy for inborn errors of cobalamin metabolism and remethylation disorders.

In addition to Dr. Charles Venditti the other round table participants included Dr. David Rosenblatt, Dr. Sally Stabler, Martyn Hooper (patient representative from the Pernicious Anemia Society), Jean-Louis Gueant, Irwin Rosenberg, Joel Mason and Ebba Nexo; all experts in the area of B12 metabolism on a research or clinical basis. It was an incredible group and the discussion was focused on the needs of the patients and short comings of our medical understanding on how to diagnose and treat these various disorders. Following the round table, the participants are currently working on an editorial for the medical journal Nutrition to be published soon.

SPECIAL EDITION, CONT'

While participating in the round table discussion was the main focus of Brittany's trip, she was also able to attend two additional days of the conference and was pleased to hear presentations from several researchers focused on cobalamin disorders. Dr. Venditti presented to the whole group (in addition to the round table) about the amazing work his team is conducting at the NIH. They have been gathering data on the improvement of cognitive and optical outcomes when patients are given high doses of Hydroxocobalamin. The effects were especially noted in children under the age of 10. He also explained an interest the team has in gene therapy for cobalamin c disorders after promising results in KO mouse models.

Dr. Jean-Louis Gueant and David Cohelo from France also presented very promising research conducted using a mouse model for Cobalamin G defects and a new small molecule that could have the potential to rescue and prevent damage done to the brain and eyes from complications related to the disorder. While other researchers and PhD students may not have been specific to cobalamin disorders, many presented very important information related to the folate and methionine pathway. A greater understanding of that biochemical pathway will only aid in future studies and advance therapeutic options for this group of disorders.

Overall it was a fruitful experience and Brittany was able to make many new connections with researchers interested in this group of disorders. Many were delighted by the community we have gathered together and the resources we have available for families.

SEPTEMBER NBS WEBINAR RECAP

We appreciated the opportunity to share what Classical Homocystinuria is, the importance of early diagnosis through NBS screening, and the changes needed to help ensure babies born with HCU are no longer missed.

A special thank you to our moderator, Dr. Mark Korson & our panelists, Dr. Marzia Pasquali & Justin Skeens

Check out the recording on our Youtube channel! https://bit.ly/3dPZaGb

EVENT ANNOUCEMENT

What are the Homocystinurias?: A Panel Perspective

October 5 at 5 PM EDT

Danae'
Classical HCU Patient

Misty
CblC Parent

HCU NETWORK AMERICA

Brittany
CblG Parent

Liz
Classical HCU Parent

Grace
MTHFR Parent

Register to watch here: https://bit.ly/3LZ2Kul

UPCOMING EVENTS

Find all events at: https://www.eventbrite.com/o/hcu-network-america-30163980100

Classical HCU Parent-Caregiver Meetup

Parents,Grandparents and Caregivers of "kids" all ages with HCU need support too!
Saturday October 8, 2022
10 am CST / 11 am EST

Classical HCU Patient Virtual Meet-up

Online meet-ups are an opportunity to for patients impacted by homocystinuria to connect one another virtually.

Sunday, October 9 2022
2 pm CST / 3 pm EST

Cobalamin Disorders with HCU Community Virtual Meet-up

Online meet-ups are an opportunity to connect patients and caregivers impacted by cobalamin disorders with elevations of homocysteine to one another virtually.

Sunday, October 23 2022
2 pm CST / 3 pm EST

EVENT ANNOUCEMENT

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

FACEBOOK LIVE CHAT

October 27 at 9 am ET

Suzanne Hollander

RD (Dietitian)

Kim Chapman

Metabolic Geneticist

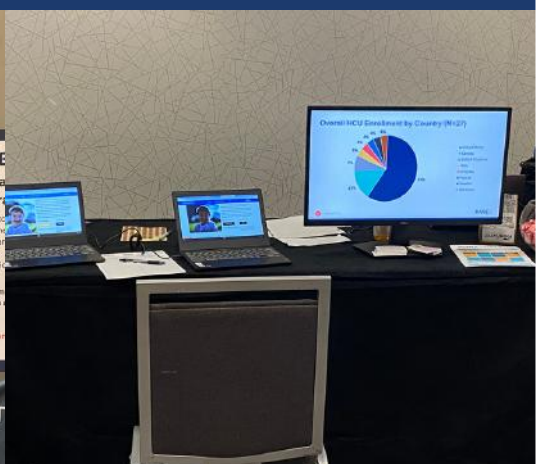
Danae' Bartke

Patient

Topic:
How the Rare-X Homocystinurias Data Collection Program Could Drive Standards of Care

HCU NETWORK AMERICA | HCU Network Australia | POWERED BY RARE X

Not a Facebook user? No problem!
You can register to watch here: https://bit.ly/3xTTS3j



EVENT ANNOUCEMENT

A KOL Webinar on Classical Homocystinuria and Pegtarviliase, Aeglea's Potential Treatment

Thursday, October 27, 2022 | 10:00 AM EDT

Hosted by Aeglea BioTherapeutics (NASDAQ: AGLE)

Join us for the Aeglea BioTherapeutics KOL Webinar featuring expert **Harvey Levy, MD (Boston Children's Hospital and Harvard Medical School)**, who will discuss the unmet medical need and current treatment landscape for patients suffering from Classical Homocystinuria, also known as cystathionine beta synthase (CBS) deficiency.

The Aeglea BioTherapeutics management team will provide an overview of their potential treatment solution, pegtarviliase, a human enzyme therapy which is currently in Phase 1/2 clinical development.

A live Q&A session will follow the formal presentations.

Register here: https://bit.ly/3UV3IeT

Featuring
Dr. Harvey Levy

Speaker Biography

Dr. Levy is Senior Physician in Medicine/Genetics at Boston Children's Hospital and Professor of Pediatrics at Harvard Medical School. He has been involved in metabolic diseases for over 50 years, including laboratory and clinical research, diagnosis, treatment and long-term follow-up as well as newborn screening (NBS). Homocystinuria has been a major involvement. It was the first metabolic disease that he encountered during his post-doc training at the Massachusetts General Hospital, and he has continued his interest in Homocystinuria ever since. He established the Metabolic Program at the Boston Children's Hospital when he moved from the Massachusetts General Hospital in 1978 and it has become one of the world's premier such programs. The program was recently named in his honor and the Director of the program is given the title of the Harvey L. Levy Chair in Metabolic Disorders. Dr. Levy has published almost 500 articles on metabolic disease and numerous chapters in classical metabolic texts as well as two books. He has received numerous awards and citations.

His interest in Homocystinuria dates to when he diagnosed the first baby identified with Homocystinuria by NBS in the United States and one of the first worldwide. He then diagnosed the older sister born before NBS when he found that she had the clinical features of Homocystinuria. He has followed this family for over 50 years, and has diagnosed, treated, and followed over 40 additional patients with Homocystinuria as well as consulting on numerous patients at other medical centers nationally and internationally.

Currently, Dr. Levy is the Principal Investigator of the Boston site for the clinical trial of an enzyme replacement therapy for Homocystinuria sponsored by Travere and is Consultant to Aeglea on their clinical trial of a human enzyme therapy for Homocystinuria.

EVENT ANNOUCEMENT

Homocysteinemias online course

November 7-18, 2022

- The course targets primarily (but not exclusively) advanced practice providers and clinicians who:
  - have a science background
  - have had no/limited formal training in genetic metabolic disease
  - are interested, or are involved, in the diagnosis/management of patients with inborn errors of metabolism

- Free of charge for physicians & trainees, nurse/nurse practitioners, physician associates, genetic counselors, dietitians; $100 corporate applicants

Learn more and sign up here: https://bit.ly/3UPLfk7

Approved for 7.0 AMA PRA Category. 1 Credits™/ 7.0 AAPA Category 1 CME credits/ 7.0 ANCC Contact Hours/ 7.0 Credits for Dieticians

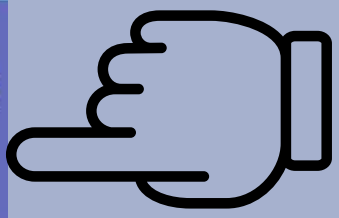
VMP, llc

Expanding Genetic Horizons

Paid study opportunity!

You're Invited!

Your feedback is important.



CLASSICAL HCU TREATMENT DEVELOPMENT

Market Research Study

We are seeking individuals to participate in a market research study discussing their experience living with classical homocystinuria and their interest in a new medical treatment that is currently being studied.

If you:

- live in the U.S., UK, Germany, or Brazil

AND

- have been diagnosed with **classical homocystinuria**

OR

are a **caregiver** for someone who has been diagnosed with classical homocystinuria

AND

- are **18 years or older**

you may qualify to participate in a Zoom interview study. All information and responses during this interview will remain confidential.

If you qualify and complete a 60-minute interview, you will be **compensated $100 for your time and participation**. Interviews are being run until **October 7.**

Interested?

To see if you qualify, please fill out the following form and a representative from ClearView will be in touch!

https://forms.office.com/r/7iqgSgP93X

We'd like to hear about your current classical HCU management plan and level of interest in a new treatment in development.

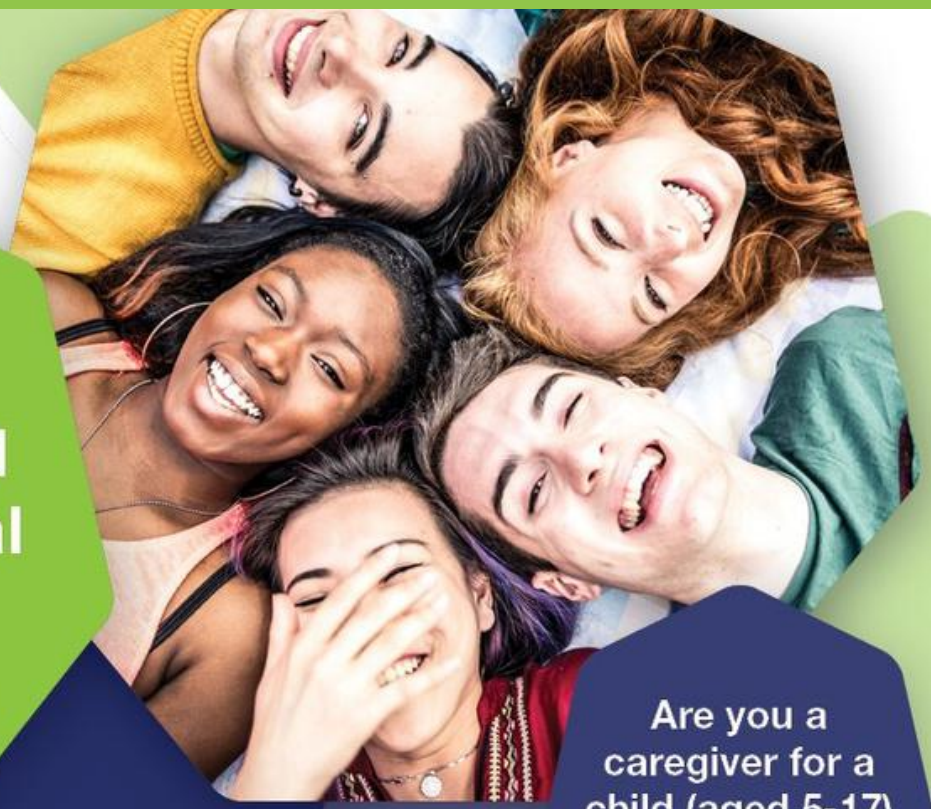
Example Topics:

- **What medical or lifestyle treatments have you been prescribed for classical HCU?**
- **How has your treatment plan changed over time?**
- **How satisfied are you with your current treatment plan?**
- **If a new treatment became available for classical HCU, what questions would you have?**
- **What factors would you consider before switching to a new treatment?**
- **What medical or lifestyle goals are you looking to achieve from a new treatment that you have not been able to achieve from your current treatments, if any?**

Please note, access to a laptop or tablet will be necessary to complete the interview as we will ask you to look over a brief description of the new treatment

Paid study opportunity!

Evidera | PPD



Patients or Caregivers Needed for a Paid Interview Classical Homocystinuria (HCU) Research Study

Are you an adult with HCU?

Are you an adolescent (aged 12-17) with HCU?

Are you a caregiver for a child (aged 5-17) with HCU?

YOU MAY QUALIFY to take part in a non-treatment research telephone/web-based teleconference interview study.

YOU WILL BE INTERVIEWED about HCU symptoms and the impact that it has on everyday life. You will also be asked about your thoughts and opinions on some questionnaires.

YOU WILL ALSO be asked to complete some questionnaires about yourself (or your child, if you are a caregiver). The interview will take place over the phone or via a web-based teleconference platform.

- This is an interview study only and does not involve any medical procedures.
- Enrolled eligible participants will receive a total of $150 (US participants) paid via gift card.
- The telephone/web-based teleconference interview can be scheduled anytime.

For more information about this study, please contact the Evidera research team.

HCUStudy.sm@evidera.com

Participate in a Homocystinuria (HCU) Research Study

This study is looking at the safety of an investigational drug, pegtarviliase, and how well it is tolerated in patients with Homocystinuria (HCU)

About The Study

- Patients will participate across the globe
- Your participation will last approximately 14 weeks to include a screening period, 4 weeks of treatment, and a follow-up period
- You and your healthcare provider will know that you are taking the study drug (Open Label)
- Some study visits may be done at your home
- Study-related expenses (travel service, reimbursement for loss of earnings, and other study-related expenses) will be provided

Interested in Participating?

Please contact Kellyn Pollard and Juana Luevano at:

GeneticsResearch@utsouthwestern.edu

at The University of Texas Southwestern Medical Center in Dallas, TX

Why Participate?

- Help advance HCU research and therapies
- Help scientists understand how pegtarviliase works*
- You will be receiving pegtarviliase, a potential treatment before it is widely available

Who Can Participate?

- 18 years of age and older
- Diagnosis of HCU due to Cystathionine β-Synthase (CBS) deficiency
- For additional eligibility criteria, please scan the QR code below, or visit: https://bit.ly/3CX7XAd

*This study may be of no benefit to you. Taking part in this study may or may not improve your health.

aeglea.com

aeglea

NOW ENROLLING: A Natural History Study on Classical Homocystinuria

Travere Therapeutics is enrolling children and adults with classical homocystinuria (HCU) in a Natural History Study. The goal is to learn more about classical HCU and course of the disease. Information gained from this study may help to improve understanding of HCU and help other patients, families, healthcare providers, and researchers to design new clinical research studies and therapies. No investigational medicine will be given to patients.

Approximately 150 patients will participate at sites in the US, Europe, and other countries around the world. The study will include three key stages (screening, enrollment, and observational follow-up) and will last approximately 6.5 years.

You (or your child) may be eligible to participate in the Natural History Study if you:

- **Have been diagnosed with HCU**
- **Are 5–65 years of age**

You (or your child) will need to meet all other study criteria to take part in the Natural History Study.

For additional information about the Natural History Study, please go to: https://www.clinicaltrials.gov/ct2/show/NCT02998710

You may be able to receive payment for time and travel when you participate in this study. Talk with your doctor and family members about joining the Natural History Study. Sites are open and currently enrolling patients.

If you have any questions, please email:

HCUConnect@labcorp.com

Visit www.hcuconnection.com for more information

MA-PE-22-0004. March 2022.

TRAVERE THERAPEUTICS

Compose

COMPOSE Study

A study that looks at how safe pegtibatinase is and how well it works in people with classical homocystinuria (HCU)

NOW ENROLLING

Travere Therapeutics has initiated a first-in-human study of pegtibatinase, a new, investigational human enzyme therapy that targets the underlying enzyme deficiency that causes HCU.

The goal of this study is to learn how safe and effective pegtibatinase is and how well it works in people with HCU at different dosage levels.

Approximately 32 subjects will participate in sites in the US. The study will include three key stages (screening, treatment, and extension) and will last approximately 150 weeks.

You (or your child) may be eligible to participate in the COMPOSE Study if you:

- **Have been diagnosed with HCU**
- **Are 12–65 years of age**

You (or your child) will need to meet all other study criteria to take part in the COMPOSE Study.

For additional information on criteria for eligibility, please go to: www.clinicaltrials.gov/show/NCT03406611

Payment for time and travel may be available to subjects who participate in this study.
To inquire about participation in the study, **please contact**:

HCUConnect@labcorp.com

TRAVERE THERAPEUTICS

MA-497. August 2021

Live better, together!

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. 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://bit.ly/3OJuF1W