The HCU Herald

Featuring...

HCU Heroes

Xander & Max from the UK

April 2024

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

HCU HEROES: XANDER & MAX FROM THE UK

Hi, I'm Sophia Tattersall, mum to Xander and Max Tattersall. We live in Barnsley, South Yorkshire in the UK.

Xander was 6 years old and Max was 3 when they were diagnosed with classical homocystinuria. Xander is now 17 years old, and Max is now 14.

Both children were born naturally with easy pregnancies. Xander was born with an enlarged kidney and underwent pyeloplasty surgery to correct this when he was 9 months old, but nothing was picked up in regard to homocystinuria at that time. Both children were also receiving early help with things like speech and language, and both already had an EHCP plan in place, which is an Education Health and Care Plan in the UK where we are from.

Xander hit all of his early milestones but then we had major problems with toilet training and his behaviour became absolutely terrible. Max was very late crawling; he was a bum shuffler, and he didn't walk until he was 2.5 years old, which is also when he started having small seizures. Eventually, they built up to the point where he was having over 150 seizures a day. He underwent an EEG and was kept in the hospital for a series of blood tests and to start epilepsy medication. He was also in status epilepticus which is one big fit. It's really dangerous, but he was conscious and able to function. When the blood tests were done his blood was really strange, almost like jelly.

HCU HEROES: XANDER & MAX FROM THE UK

A week later on 6th August 2013 whilst me and my husband Lee were getting our heads around the epilepsy diagnosis, we received a phone call that Max had been diagnosed with a metabolic disorder and could we attend Sheffield Childrens Hospital on Friday 9th August, and to bring Xander too as he may also have the same disease. It was then that we met our Doctor, Professor Yap, and our metabolic team, and everything was explained to us.

Xander & Max with Professor Yap

I found it really hard to understand the science but decided to concentrate on what the dieticians were telling me: that we had something that could potentially make things better for the boys - the low protein diet. And if I could make a difference for them by producing delicious food that they would like, that's what I would do.

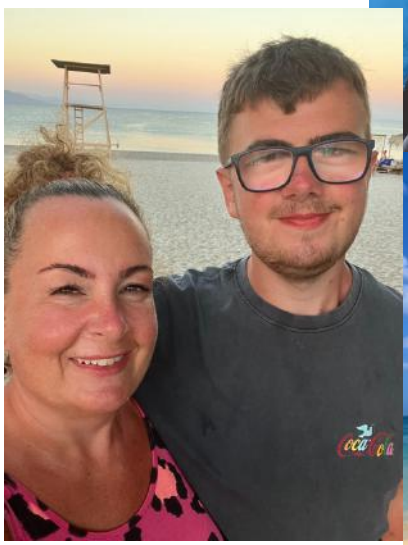
It was a really hard time for us getting our heads around everything and it certainly has not been a straightforward journey. There have been many bumps in the road, mainly with Max, as he likes to throw us medical curveballs! Xander settled straight into the diet and drank his supplements straight away. Initially, Max did too, until everything settled down and he started spitting his epilepsy meds out and refused his supplement.

HCU HEROES: XANDER & MAX FROM THE UK

Max ended up having a gastro tube fitted into his stomach in June 2014, and although it was my worst nightmare and I felt like an absolute failure, it was the right thing to do and has helped him many times when he has been seriously ill.

Max has the additional complication of Lennox-Gastaut Syndrome, which is a rare epilepsy syndrome. When he hit puberty, his epilepsy was dire until March 2023 when he started on cannabis oil, and I have to say it has changed his life. He takes this alongside other epilepsy medications. Max is nonspeaking* and attends a special school. Xander attends a specialist speech and language school, which he has to travel to on a daily basis and it is over an hour away from where we live.

Before the boys were diagnosed, I had a gut feeling that it was something medical that was wrong - always trust your gut. Homocystinuria and epilepsy haven't held us back; we have traveled, and we are going on a cruise with the boys this year.



> Homocystinuria and epilepsy haven't held us back; we have traveled, and we are going on a cruise with the boys this year.

*see notes about the term 'nonspeaking on the following page

HCU HEROES:
XANDER & MAX FROM THE UK

My hope for the future is that an enzyme therapy is developed that will mean that people with HCU will be able to eat a normal diet, and I feel that that anything less than this is not worth it – both my boys have excellent control of their condition with the diet and don't take betaine.

Always remember calories and hydration are key as well as hitting your protein target for the day. I can't believe that we are now nearly 11 years down the line, and I am so proud of us all because it hasn't been an easy ride.

Notes from Sophia:

nonspeaking vs **nonverbal**

Why do we say 'nonspeaking' and not 'nonverbal'?

Max makes a lot of noises but does not speak, therefore, he is ***nonspeaking***.

What does this mean to us?

Max can get easily frustrated when he wants to express a choice or something overstimulates him that he does not like, and he reacts through behaviour, which can sometimes be challenging.

He communicates through behaviour, which those close to him understand.

It also means that we, his parents, are his voice.

Click here to read Xander & Max's story on our website

We need Patient Stories!

BY SHARING YOUR STORY, YOU CAN....

- Spread awareness
- Help others to feel connected
- Share insight beyond our patient community of what it's like to live with homocystinuria
- Provide hope to other families

Ready to share your story?

Send us a message on social media or email us!

info@hcunetworkamerica.org

HCU NETWORK AMERICA

JOIN OUR FUNDRAISING TEAM

We are looking for new community members to join our fundraising team!

Help create, organize and support new and existing fundraising ideas.

These vital funds help support our outreach, programs and research!

To join, email Dbartke@hcunetworkamerica.org

UPCOMING EVENTS

HELP NAVIGATE THE MAZE

START

HOPE

FINISH

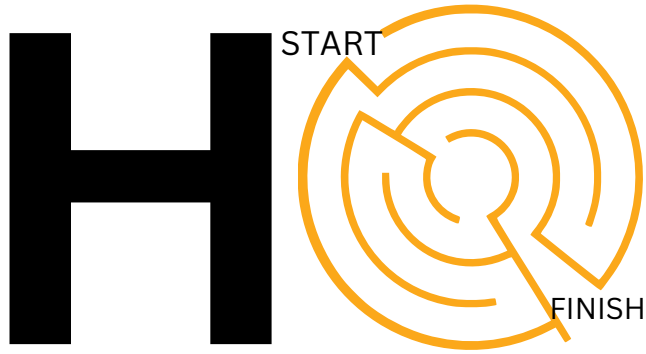
CONNECTS US

VIRTUAL RAFFLE: MAY 10TH-18TH

WINNERS ANNOUNCED VIA LIVE STREAM: MAY 18TH 4 PM ET

Help Navigate the Maze

World Homocystinuria Awareness Day | May 18, 2024



CONNECTS US

VIRTUAL RAFFLE:
MAY 10TH-18TH

LIVE STREAM:
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HCU NETWORK AMERICA

$15,000 Match for all funds raised!

All funds to benefit HCU Network America's Community Outreach and Education Programs

Registration:

TICKETSIGNUP.IO/TICKETEVENT/HCUCONFERENCE

MOVING MOUNTAINS

Classical HCU | Cobalamin Disorders | Severe MTHFR

Patient-Family Conference

Aurora, Colorado
June 29-30, 2024

HCU NETWORK AMERICA

- KEYNOTE FOCUSED ON MENTAL HEALTH
- AGING WITH THE HOMOCYSTINURIAS PANEL
- BREAKOUT SESSIONS
- DIAGNOSTIC TOOLS AND NEWBORN SCREENING
- RESEARCH UPDATES
- HCU HERO AWARD RECEPTION

UPCOMING EVENTS

HCU NETWORK AMERICA presents

ASK ME ANYTHING with flok

Mon 4/22 7pm ET

- How can I track my health with the flok app?
- What resources does flok provide for the HCU Community?
- What's flok Family Camp all about?
- Other Questions?

Join HCU Network America as we meet with leaders of flok *(formerly PKU News)*!

We'll talk about flok programs like their Family Camp, learn about the flok app, and take any questions you may have for the flok team.

We hope you'll join us! REGISTER

flok Together: Camp and App with Us!

flok is advancing research and care for people with Classical Homocystinuria. **The flok app** helps you track how you eat, feel, move, and play to offer new insights into our conditions.

Attend our in-person **flok Family Camps** in Oregon and New Hampshire! Our metabolic community members, family, and friends are all welcome for 3 days of fun, learning, and social connection.

flok FAMILY CAMP

Visit flok.org for more information on the flok app and Family Camp registration

flok Adult Retreat

May 28-30, 2024
Washington Family Ranch
Currant Creek Lodge
Antelope, Oregon

flok is excited to host our first adult retreat. This 2-day event is inspired by adults for adults! Our primary goal is to provide a relaxing and enjoyable opportunity for connection, learning, and fun for community members 21+ with Classical HCU, MSUD, an Organic Acidemia, PKU, Tyrosinemia, or a Urea Cycle Disorder.

Cost
$20

Registration
Contact **kristen@flok.org** for additional information and registration details

Questions?
Contact **camp@flok.org**

Activities

In addition to taking in the breathtaking views, participants will have access to hiking trails, sand volleyball, an 18-hole disc golf course, and an 88,000 square foot Sports Center with basketball, indoor volleyball, dodgeball, foosball, octaball, ping pong, billiards, climbing walls, a skate park and workout center. Participants will also have time for group discussion, low-protein cooking, and space to relax and connect with old and new friends.

Housing

Our reserved lodge has 13 bedrooms, each equipped with bunk beds to comfortably accommodate up to 30 adults. Currant Creek is not just a place to stay; it offers a spacious great room and open kitchen, providing the ideal setting for fostering connections and creating memorable experiences.

Accessibility

If you have special housing needs, let us know during registration or contact **camp@flok.org**. All walkways around camp are paved and all main buildings are designed with accessibility in mind.

Medical Accomodations

Your health and safety are important to us. We will have an onsite OR certified medical professionals (MD, RN, or EMP-P) to provide immediate assistance. However, it is important to know the St. Charles Madras Hospital is 50 miles from camp and there is a 90-minute EMS response time. Washington Family Ranch has a Life Flight contract in place. If you have questions or concerns, please reach out to us at **camp@flok.org**.

Garlic Butter Jackfruit

Makes about 3 servings **| 1 serving = 2 oz | 1.1 grams protein per serving**

Ingredients:

- 1 20-oz. can Jackfruit, canned, drained, rinsed and seeds removed
- 1 tsp Paprika
- 1 tsp Garlic Powder
- 1 tsp Onion Powder
- 1 tsp Salt
- 1/2 tsp Black Pepper
- 1 TBSP Olive Oil
- 2 tsp Lemon Juice
- 3 TBSP Butter
- 2 clove(s) Garlic, minced
- 2 TBSP Lemon Juice

Directions:

1. Preheat oven to 350 degrees and line a baking sheet with foil. Spray the foil with cooking spray.

2. Squeeze excess liquid from the jackfruit. Put it into a medium bowl and toss with olive oil, garlic powder, paprika, onion powder, salt, pepper, and lemon juice. Mix to combine and put on prepared baking sheet in a single layer. Roast in the oven for 10 minutes. Turn the pieces over and roast for another 10 minutes.

3. While the jackfruit is roasting, make the sauce. Melt the butter in a small skillet over medium heat. Once the butter is hot, add the garlic and stir until aromatic, at least 2 minutes. Add the lemon juice and cook for 5 minutes. Remove the jackfruit and toss in the butter sauce. Serve hot with rice, potatoes, or roasted veggies.

What's your dream?

MAKE IT HAPPEN!

#RAREis Scholarship Fund
Powered by the EveryLife Foundation

The #RAREis Scholarship Fund enriches the lives of adults living with rare diseases by providing support for their educational pursuits.

Applications open March 18 - April 22, 2024 at RareScholarship.org

What is the scholarship amount?

- One-time awards of $5,000 each will be granted to up to 88 recipients for the Fall 2024 semester.

Who can apply?

- Anyone diagnosed by a physician as having any form of rare disease regardless of treatment status. A disease is defined as rare when it affects fewer than 200,000 people in the United States. Undiagnosed diseases qualify.
- Anyone age 17 or older and who are residents of the United States.
- Applicants must plan to be enrolled in an accredited educational program for the Fall 2024 semester. Educational programs include full-time or part-time undergraduate or graduate studies, vocational-technical or trade school, or accredited classes. No minimum number of classes or credits are required.
- Students are eligible to apply and receive the scholarship no more than four times.

How do I apply?

- Visit rarescholarship.org and click 'apply now' at the top
- You will be taken to Scholarship America's application portal. Enter your information
- Answer the 1-page essay question about your rare disease journey
- Upload documents: your most recent transcript and Diagnosis Verification Form
- Review & submit your application by April 22, 2024 at 2:00 PM ET

EVERYLIFE FOUNDATION FOR RARE DISEASES | 15th ANNIVERSARY

#RAREis Scholarship Fund
Powered by the EveryLife Foundation

AMGEN

Incase you missed it...

The HC&U Podcast is back!!!

HC&U is a podcast about Homocystinuria, sponsored by HCU Network America and hosted by Ben & Lindsey.

Meet your hosts!

Welcome to the HC&U Podcast! We are Ben and Lindsey, your hosts. We are so excited to be starting this as extra resources for the Homocystinuria community. We hope you like our content!

To Listen:
https://hcunetworkamerica.org/hcu-podcast/
or click below on your favorite option!

LISTEN ON Spotify

Listen on Apple Podcasts

LISTEN ON iHeartRADIO

Listen on amazon music

The latest episode

In this episode, Ben welcomes Jamela Gutierrez to the table! Jamela, an adult patient with HCU and a medical professional, will share her insights and experiences with the low-protein diet, medical formula, and much more!

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

Spotlight on Medical Diets!

Thank you for taking the Interventional or Medical Diets Survey. Here are the findings!

34 Classical HCU participants responded to the survey

79% report having been prescribed a low-protein diet

29% report taking medical formula

26 Classical HCU patients responded to the medications survey

- 73% take Betaine
- 46% take folic acid or folate
- 42% take B6
- 35% take B12

Haven't taken the Interventional or Medical Diets Survey yet?
Make your voice heard! Head to https://homocystinuria.rare-x.org/

CANPKU+ CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

Sign our NBS Screening Petition!

In this petition, we call for state newborn screening labs to **revise screening protocols for Classical Homocystinuria** to *ensure fewer false negative screening results and delayed diagnoses.*

Click here to sign the petition!

Get your kit!

Our FREE Customizable Kits are here!

Request yours today!

At HCU Network America, we believe that one of the most important steps to empowering patients and caregivers is giving them the support and tools needed to succeed! We know that a new diagnosis can be overwhelming and riddled with concerns and questions. To us, one way to combat those feelings, and give you the confidence you need, is by providing you with one-on-one support, educational resources, and practical tools, such as scales, cooler bags, and more! Our request for a kit survey allows you the opportunity to request a one-on-one introductory call (with more opportunities to connect), and then a customized kit to the patient's needs. Don't want a call or a Zoom? That's fine too - we are happy to send you the customized kit.

Request your kit now - **https://www.surveymonkey.com/r/HCUKitSurvey**

**Kits can only be sent to patients in the continental US. However, we are happy to connect virtually and share the educational materials with you via weblinks*!

UPDATE

Now includes funding assistance for medical formula AND low protein foods!

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

What is the purpose of this program?

Having a rare disease is difficult. Adding in the complex care required to treat or manage that disease and figuring out how to pay for it makes a rare diagnosis even harder.

NORD's Classical HCU Medical Assistance Program offers eligible individuals diagnosed with Classical Homocystinuria financial support to pay for the low protein foods and physician prescribed medical formulas necessary in managing this HCU diagnosis.

NORD's HCU Medical Assistance Program opened thanks to a generous donation from the HCU Network America.

MEDICAL ASSISTANCE

Who is eligible to apply?

This program is designed to help patients who:

- Have a diagnosis of Classical Homocystinuria.
- Are a United States citizen or U.S. resident of six (6) months or greater with evidence of residency such as a utility bill showing the patient's name and address.
- Meet the program's financial eligibility criteria.

What is the application process?

Patients may be referred to the program by their health care provider, their case managers, or they may self-refer.

A NORD Patient Services Representative will guide the applicant through the application process and verify eligibility for inclusion in the HCU Program.

Awards are based on meeting eligibility criteria, funding availability, and are made on a first-come, first serve basis.

NORD is Here for You

NORD, a 501(c)(3) organization, is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.

NORD was founded by families struggling to obtain access to treatments and whose advocacy for change led to the passage of the Orphan Drug Act in 1983. NORD assists eligible patients (those with medical and financial needs) in affording the treatments and medical services their healthcare professionals have prescribed.

Alone we are rare. Together we are strong.®

How do I get more information and apply?

Contact NORD's Classical HCU Medical Assistance Program

Monday-Thursday 8:30am – 7:00pm ET
Friday 8:30 am – 6:00pm ET

203-616-4327

203-635-4163

hcu@rarediseases.org

US MAIL to: NORD
Attention: HCU Program
55 Kenosia Avenue
Danbury, CT 06810

What assistance does NORD provide?

- The Classical HCU Medical Assistance Program assists eligible individuals with out-of-pocket costs to purchase low protein foods and physician prescribed medical formulas. Individuals approved for assistance in this program will be issued a PEX card. The PEX card is a prepaid expense card to be used for the purchase of low protein foods or physician prescribed medical formulas only.
- *Upon receipt of the card, the cardholder will contact NORD to request card activation. The card will be funded based on program award caps set for the program (this cap will be discussed with individuals upon enrollment in the program..*
  - *> It is necessary for the individual to submit receipts on a monthly basis evidencing card utilization for the purchase of low protein foods or medical formulas for the previous month.*
  - *> Funds will not be added to the card until the previous month's receipts have been received by NORD.*
  - *> The card may only be utilized for the purchase of low protein foods up to the monthly program limit.*

Once a patient is accepted into the assistance program(s) how long are they eligible?

Awards are issued for a calendar year.

Patients are encouraged to reapply annually if continued assistance is needed.

What happens if an applicant does not meet the criteria of the Electronic Income Verification?

The NORD Patient Services Representative will offer to e-mail, fax, or mail the brief program application and disclosure forms to the patient. The applicant may then complete the application, sign the disclosure form, provide the appropriate financial documentation to verify financial need, and return them via fax, email, or USPS mail.

How does NORD demonstrate compliance with regulations required of charities?

- NORD independently designs its patient assistance programs based on the needs of specific patient communities.
- No pharmaceutical company or donor controls or influences our programs.
- Our patient assistance decisions are based on consistently applied financial eligibility criteria and diagnosis only.
- Patients have their choice of health care provider, treatment and treatment location, and can make changes at any time.
- Patients' privacy and well-being are priorities at NORD. We do not share or provide patient names or data with donors, nor do we disclose or identify donors to patients. Patients are able to make the choices that are best for them because NORD's assistance covers all FDA-approved products available for a diagnosis. Our programs also help with more than medication: patients can use their funds to pay for other physician prescribed services related to their diagnosis, such as laboratory and diagnostic testing, physical and occupational therapy, durable medical and adaptive equipment, and travel to medical appointments.

NORD®

rarediseases.org

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://www.surveymonkey.com/r/HCUContact**

FOLLOW US