The HCU Herald

Featuring...

HCU Hero
Jacob from New Jersey

December 2022

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

HCU HERO: JACOB FROM NEW JERSEY

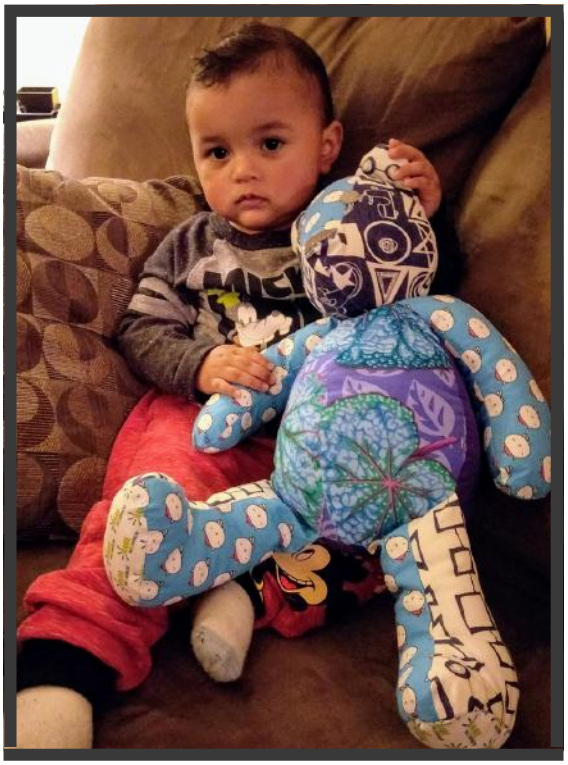
Our son Jacob was born on September 22, 2017. He was long awaited by his older brother and us. He was absolutely precious and our perfect little bundle of joy. Then, on September 30, our lives were turned upside down by a phone call from the State. After confirming that I was Jacob's mother, they asked if the pediatrician had reached out to me regarding Jacob's Newborn Screening results. I told them that I had not received a phone call and asked if everything was ok. In turn, they said they could not give me any information, but I should reach out to our pediatrician. I didn't quite understand why the State was calling me regarding test results for our son if they could not discuss the results with me.

Just as I was hanging up the phone, the pediatrician's office called to schedule us to go in right away. When we got to the office, Jacob's doctor let us know that his methionine levels were high, at 107, and that he would need to be retested to confirm because he may have a metabolic disorder called Homocystinuria. We had never heard of this disorder and asked her if she could explain. She told us she had only heard of this condition once before as a resident doctor and could not provide much information. Needless to say, we left the office feeling scared, confused, and defeated as we did not know where to turn.

Jacob was retested on October 3, 2017, and his methionine levels were even higher at 703. I do not remember being told that Jacob's levels were so high. In fact, during a recent conversation with our genetic counselor, she said she may not have given us the number to avoid scaring us even more. (It is important to note that in New Jersey, the methionine cutoff for a positive result on the Newborn Screening test is 80 to 110.) But honestly, at that moment, we would not have known this number was so important. We will be forever grateful that our metabolic team began treating Jacob to get his levels down immediately. From day one, our genetic counselor has been a Godsend because she has always been very informative and helpful.

> **We realized that Jacob's early diagnosis was a blessing.**

After Jacob's diagnosis, we continued to do our own research, hoping to find anything that would give us more insight into Homocystinuria. We still felt worried and needed answers to so many questions. I came across the HCU Network America website and there I learned they were soon having their first-ever HCU Conference. I reached out to Danae to inquire about the conference and speak about Jacob. She answered many of my questions and encouraged me to bring Jacob along. The conference was such an eye-opener for us! We learned so much from the experts and met some amazing people. We realized that Jacob's early diagnosis was a blessing.



Jacob is an intelligent, vibrant, strong-willed five-year-old. He is managing his Homocystinuria well. His treatment consists of Cystadane, B6, B12, Folic Acid, L-Cystine, a low protein diet, and his supplemental formula. When we were originally told that Jacob would need to be on a low-protein diet for the rest of his life, we were concerned and thought about all the things he would miss out on. However, today, I feel that his diet has allowed us to eat healthier as a family. We have adapted and learned how to make substitutions with our meals so that Jacob does not have to feel as if he eats very differently from us. We have been very fortunate to have family members and friends that are always happy to make adjustments for Jacob during gatherings.

During these five short years, we have been Jacob's advocates in so many ways. On numerous occasions, we have had to explain Jacob's disorder, his diet and the medications he cannot take due to his condition, to doctors, nurses, and teachers. We will continue to be his number one advocates for as long as we need to.

"...his diet has allowed us to eat healthier as a family. We have adapted and learned how to make substitutions with our meals so that Jacob does not have to feel as if he eats very differently from us."

To all of the parents out there that have recently received this diagnosis, please know that you are not alone. It is ok to be scared, and it is ok to be saddened but know that with time you will develop the strength you need to move forward. Do not be afraid to reach out to The HCU Community. This community did wonders for our family. It is full of wonderful people that are always willing to lend a helping hand.

This HCU Hero story was written with love by Liezza, mom of Jacob.

LO PRO HOLIDAY DESSERT

Chocolate Celebration Cake

Serves 12

ingredients

- 1-15.25 oz package chocolate cake mix
- 4 tablespoons instant chocolate pudding mix
- 12 ounces cola
- 3 ounces water
- 1-16 oz container frosting flavor of choice

NOTES:

~

FOR CUPCAKES, FILL EACH TIN HALFWAY. RECIPE WILL MAKE 24 CUPCAKES. PROTEIN AND AMINO ACID VALUES WILL BE HALF.

~

SIZE OF CAKE WILL VARY BASED ON PAN USED. REMEMBER TO DIVIDE EVENLY INTO 12 SERVINGS.

directions

- Heat oven to 350ºF.
- Using the instructions on the cake mix box as a guide, select your baking pan, then grease and flour it.
- Combine cake mix, pudding mix, cola and water in a large bowl. Stir with a spoon or beat with a mixer for 2 minutes.
- Bake according to times on cake mix package based on your pan used. Cake is finished when a toothpick inserted comes out clean.
- Cool cake completely before frosting. Divide cake into 12 equal servings.

KCAL: 340 PROTEIN: 1 G ARGININE: 88 MG METHIONINE: 31
DETERMINED BY NUTRITIONIST PRO.

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: Pumpkin Bread & Banana Slices
Lunch: Grilled Cheese & Butternut Squash Soup
Dinner: Potato Pizza & Veggies

T

Breakfast: Eggz, Avocado & Fruit
Lunch: Pasta Salad
Dinner: Tacos

W

Breakfast: Cinnamon Buns & Grapes
Lunch: Vegetable Chili
Dinner: Fried Cauliflower & Mashed potatoes

T

Breakfast: Cereal & Fruit
Lunch: Biscoff & Jelly Sandwich & Pretezel Sticks
Dinner: General Tso Stir Fry

F

Breakfast: Breakfast bar & yogurt
Lunch: Veggie Meatball Sandwich
Dinner: Jackfruit Kabobs

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.

Make it LO PRO

aeglea

@makeitlopro

Make It Lo Pro

Looking for some new (and easy!) low protein meal inspiration?

Check out our "Make it Lo Pro" video series!

The best part? All ingredients can be found at your local grocery store!

Lo Pro at Home: Snack board

Lo Pro at Home: Potluck enchiladas

Lo Pro at Home: Sweet potato bacon

VIRTUAL FUNDRAISER

EAT PANDA EXPRESS, SUPPORT HCU NETWORK AMERICA!

28% OF SALES WILL GO TO HCU NETWORK AMERICA!

When: Friday, December 02, 2022

Place your online order for pickup or delivery on Friday, December 2!

Where: Panda Express locations nationwide

How: Online orders only

*Apply code **911897** in the Fundraiser Code box during online checkout at www.pandaexpress.com or via App*

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

MEDICAL ASSISTANCE

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 necessary in managing this HCU diagnosis.

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

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?

NORD's program can assist eligible individuals with the expense of purchasing low protein foods:

- The Classical HCU Medical Assistance Program assists eligible individuals with out-of-pocket costs to purchase low protein foods. 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 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 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.

Program assistance is dependent on funding availability.

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

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

OUR MATCHING GIFT IS BACK - AND BIGGER THAN EVER!!!

That's right, you heard us right! Thanks to two anonymous donors, **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. 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 letter on next two pages, or you can print it from [here]

15 S. Mallory Ave
Batavia, IL 60510

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

Dear, First Name

The past two years have been a struggle for many. The year of 2022 feels oddly like a new beginning, though we recognize that challenges remain that affect your lives. We sincerely hope you have been able to successfully manage these challenges.

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 must balance all the variables in their lives that affect their health and that 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 was born in the summer of 2020. She was our second daughter, and we were thrilled to have a new addition to our family. We received a call after only one week that our daughter's newborn screening was abnormal. We were assured that it was most likely nothing, but we needed to do another screening to be sure. The second screening was abnormal as well. This time, we needed to go to our nearest metabolic clinic 4 hours away to test her levels, do a genetic test, and meet with the geneticist to learn about Homocystinuria and available treatments. The genetic test later confirmed a diagnosis of Homocystinuria due to severe MTHFR deficiency. Her initial Homocysteine level was 295 (normal levels are below 15), and her Methionine was 3 (normal levels are over 20). Although our daughter's condition was caught early, she didn't begin treatment until three weeks of age. During those first three weeks, our daughter didn't gain any weight and she slept more than normal. I initially thought she was a great baby who loved to sleep, but I later realized it was due to her condition. Within a few days of starting treatment, she started becoming more alert. At that time, I noticed it was difficult for her to focus her eyes. When she tried to look at me, her eyes would uncontrollably roll up. We brought her to the local ER, and the doctor believed that our daughter was having seizures. The metabolic specialists arranged for our daughter to be flown to them so that she could be monitored through the weekend. Thankfully, all tests came back normal, and for the first time since she was born 3 ½ weeks earlier, she was actually able to look and stare at me. After only one week of treatment, she gained more than a pound. For the first 7 months, she was consistently behind developmentally by 1 month. It was as if she completely "lost" that time before treatment was started. It was also months before her head size ever made it onto the growth chart. Our daughter is now two, and she is doing better than we ever expected. Besides a speech delay, she is developing like any other two-year-old. She is incredibly smart and the happiest child. Newborn screening and early treatment saved her life!*

Since HCU Network America was available, this family was able to get the support they needed at one of the most devastating times in their lives. 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 with toolkits which are filled with helpful tips and guidelines for living with HCU.

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

- Awarded four research grants for improved therapies and one for better newborn screening.
- Held our third patient/family-expert conference
- Launched a low-protein foods assistance fund
- Provided a consultant with experience in medical insurance to fight for coverage for medications and food at no cost to HCU patients
- Supported community meetups and monthly community newsletter
- Enrolled 42 patients in our Homocystinuria Data Collection program and 23 in our biobank repository
- Advocated for improved newborn screening which led to several states adopting changes

Our goal this year is to raise $60,000. Thanks to an anonymous donor, any funds you donate up to December 31, 2022 will be matched up to $30,000. Please consider a donation now to HCU Network America. If you personally know a patient with HCU, you can donate in their honor. Ask your families and friends to help. 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 look forward to 2023!

Thank you,

Danae' Bartke

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
  Donor's name and HCU patient's name listed on website

4 ways to donate:

1. Use the enclosed slip and envelope
2. Go to https://hcunetworkamerica.org/donate
3. Text HCU2021 to 44-321
4. Scan the QR Code

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?

GET YOUR HCU GEAR!

Looking for a last-minute gift or stocking stuffer?

Want to raise awareness while also supporting the HCU community?

We've got you covered!

HCU Awareness Earrings

$15 *Includes Shipping

To order earrings, send payment via PayPal @gtalbert90

Include your shipping address

If you have any questions, email Grace Talbert at Grace.anderson90@gmail.com

WEAR YOUR AWARENESS

this holiday season

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

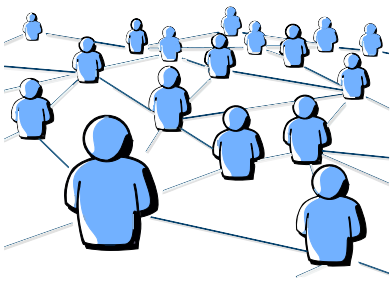
RESEARCH

Ever wondered if that issue that you've experienced is related to HCU?

(Dental, skeletal/connective tissue, heart, mental/emotional/behavioral health, etc)

There's only one way to find out for sure!

By sharing our collective experiences, we can help researchers and our providers to better understand ALL of the different manifestations of the disease.

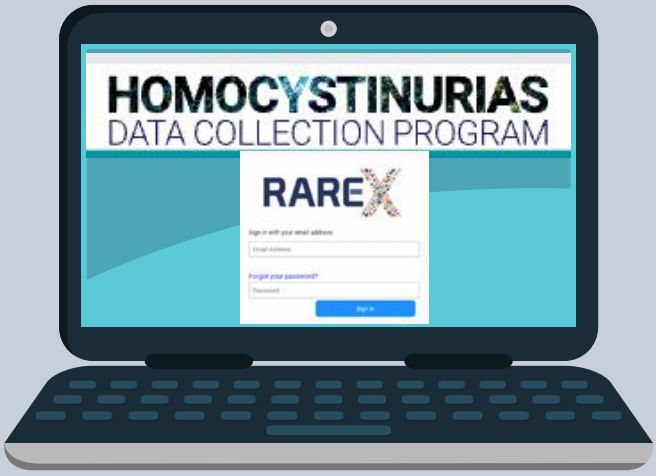


Did you know?

The experiences that we share on Rare-X will help to shape our treatment guidelines! Let's make sure that our experiences are well-represented!

REGISTER NOW!

https://homocystinuria.rare-x.org/



RESEARCH

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

FACEBOOK LIVE CHATS

- what's YOUR why?
- The Low Protein Diet
- Health and Development
- Mental Health
- Quality of Life
- Eyes and Vision Issues

Catch all of the replays here! bit.ly/3Vk2A4x

EVENT ANNOUCEMENT

Inaugural Havey Levy Newborn Screening Symposium

Dr. Harvey Levy is an internationally renowned leader in Newborn Screening (NBS) with a prolific career in inborn errors of Metabolism (IEMs) that spans decades and includes collaborations with Dr. Robert Guthrie and Dr. Charles Scriver, to lay the foundation for NBS, as the most important Public Health initiative in the United States. This inaugural symposium is an all-day virtual educational event celebrating his pioneering efforts for NBS. The symposium focuses on the past, present, and future of NBS and metabolic disorders. This annual event features international and national speakers who have expert knowledge in the field of NBS. Our speakers will be discussing the current state of NBS and its future direction with the ongoing development of therapies for IEMs.

Symposium Details

Dr. Olaf Bodamer, Dr. Walla Al-Hertani, and Alba Savinon invite you to attend the inaugural Harvey Levy Newborn Screening symposium, which will be held virtually on Thursday, December 8, 2022. Click here to RSVP, registration is free.

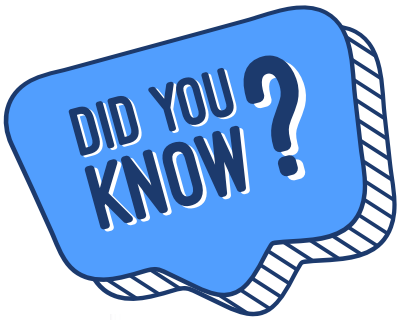
(8:15 am - 5:00 pm ET)

Click here
to join webinar

PROGRAM HOSTS

Boston Children's Hospital
Genetics and Genomics

Boston Children's Hospital
NORD Center of Excellence
for Rare Disorders



Dr. Levy developed a specific interest in homocystinuria during his fellowship when he identified the first case diagnosed from newborn screening. Shortly thereafter he and Dr. Harvey Mudd discovered the first example of increased homocysteine due to a non-homocystinuric disorder, now known as cobalamin C defect. This was also the first example of a human disorder of vitamin B12 metabolism. Disorders of sulfur metabolism have continued to be a major interest of Dr. Levy throughout his career. He has authored over 400 medical articles and book chapters on metabolic disorders, including many on the homocystinemias, and has received a number of national and international awards for his achievements.

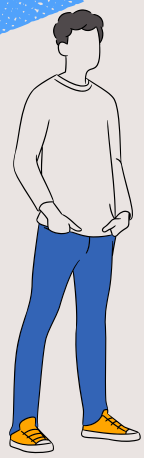
Currently, Dr. Levy and Boston Children's are in charge of one of the six U.S. sites that is participating in the Natural History Study of Cystathionine Beta-synthase Deficiency Homocystinuria (National Clinical Trial number NCT02998710) and subsequently an ongoing clinical trial of enzyme replacement therapy for homocystinuria, which will help improve our understanding of the disease course and clinical outcomes of homocystinuria and aid in the development of better treatments for this devastating disease.

NEW RESOURCE

Our *Transition to Adulthood* Guide is here!

CHECK IT OUT

#ADULTING



INTRODUCTION

THE TRANSITION TO ADULTHOOD JOURNEY

Thanks to advances in research, people with homocystinuria are living healthier and more active lives than ever before—juggling busy schedules, careers and family. If they aren't doing these things, that's okay too; life doesn't always look the same for everyone.

This guide is intended to help people with homocystinuria gain the knowledge and skills they need as they grow up to more independently manage their condition. Our flexible toolset is designed for people with homocystinuria to use with their caregivers and care teams to support the transition from childhood to adulthood.

HOW TO USE THIS GUIDE

- Chapters in this guide cover the basics, as well as medical, social, financial and legal aspects.
- The horizontal bar at the top of the chart lists several categories for performing the task, whether an individual might need help with the task, can do it on their own or will learn to do it on their own, for example.
- The code in the left-hand column indicates variations of homocystinuria.
- As you go through these worksheets, review each task and click the box that indicates whether you need help with certain tasks or can do them independently.

OVERALL PROJECT GOALS

- Support families' ability to have a transition plan.
- Create understanding that transition plans are fluid and individual.
- Understand that the Transition Program is not definitive start/end (for development), it will evolve with research, treatments & resources.

OBJECTIVES

- Each aspect of transition is inclusive to all abilities and all types of homocystinuria.
- Create a visual roadmap with each section for families to navigate the process of transition.

Download the full guide here: https://bit.ly/HCU-TTA-Assessment

NEW RESOURCE

¡Ahora disponible en español!

RETOMAR LOS CUIDADOS DE SALUD

NUNCA ES DEMASIADO TARDE PARA VOLVER AL CAMINO SALUDABLE

HCU NETWORK AMERICA

To download the full guide, visit: https://bit.ly/HCUBTC-SP

NEW RESOURCE

Educator's Guide - Cobalamin G & E

¡Ahora disponible en español!

Guía de trastornos de la metilcobalamina:

Cobalamina G y Cobalamina E

Maestros

¿Qué son la Cobalamina G y E?

Estas son condiciones genéticas raras que ocurren cuando el cuerpo no puede procesar ciertos aminoácidos (bloques de construcción de proteínas) adecuadamente. Provocan una acumulación de homocisteína (HCY) y una deficiencia del aminoácido metionina. Estos también se denominan trastornos de la metilcobalamina.

¿Qué son los trastornos de la metilcobalamina?

CblG y CblE son dos trastornos genéticos raros que ocurren cuando el cuerpo no puede procesar la vitamina B12 en metilcobalamina, que se necesita para sintetizar metionina, un componente importante para el cuerpo. CblG y CblE son formas de homocistinurias (HCU). Los niños con CblG/CblE tienen niveles elevados de homocisteína y metionina baja en la sangre.

¿Qué sucede?

Normalmente, ocurre un proceso metabólico en el cuerpo donde la homocisteína (HCY) se convierte en otro aminoácido, metionina. Una mutación genética impide este proceso, lo que da como resultado una acumulación peligrosa de HCY. Se necesitan suplementos y medicamentos para ayudar en este proceso y mantener niveles relativamente normales de HCY y metionina. Vitamina B12 (a veces por inyección) y otros medicamentos son necesarios para reducir el nivel de HCY, aumentar la metionina, y evitar daño. Los niveles altos de HCY y bajos de metionina son perjudiciales para los ojos, los huesos, los vasos sanguíneos y el sistema nervioso.

Los niveles altos de HCY pueden causar:

- Dificultades de Alimentación
- Problemas con la visión
- Convulsiones
- Falta de coordinación y tono muscular
- Problemas de movimiento
- Retraso en el desarrollo
- Discapacidad intelectual
- Microcefalia
- Atrofia Cerebral
- Desórdenes neurológicos
- Coágulos de sangre o derrame cerebral

¿Cómo puedo ayudar?

Enseñe a su estudiante como lo haría con cualquier otra persona. Los niveles de HCY pueden fluctuar. Su estudiante con CblG o CoblE puede necesitar más tiempo o atención para seguir el ritmo de la clase.

Ayude a asegurar que su estudiante tome sus medicamentos. Presión social puede hacer que los niños sean difíciles de tomar medicación, o evitarla.

Comuníquese con los padres y haga preguntas. Dado que pasa una buena cantidad de tiempo con su estudiante, puede ser el primero en notar problemas relacionados con CblG o CblE. El manejo exitoso de HCU dependerá de la comunicación entre los padres y el personal de la escuela.

Posibles efectos secundarios de los medicamentos

- Moretones
- Enrojecimiento circular en el lugar de la inyección
- Malestar de estómago
- Olor corporal/aliento desagradable (a menudo descrito como a pescado)
- Náusea

Consejos útiles

- Trátelo igual que a sus otros estudiantes. Su estudiante no está enfermo y no debe ser tratado como tal. Si sigue su régimen de medicación, puede tener tanto éxito como sus compañeros de clase.
- No permita que CblG o CblE definan a su estudiante. Establecer un sentido de sí mismo fuera de su trastorno es una parte crucial de la autoaceptación.
- Los pacientes no pueden sentir cuando sus niveles de HCY son altos (a diferencia de un diabético que puede sentir cuando su insulina es baja/alta). Se requieren extracciones de sangre periódicas para controlar la medicación y mitigar los efectos negativos. Los síntomas generalmente ocurren después de un período prolongado de niveles altos de homocisteína. Los niveles aumentan gradualmente, no aumentan repentinamente.
- Las personas con CblG y CblE pueden sufrir accidentes cerebrovasculares. Si le preocupa que su estudiante esté teniendo un derrame cerebral (los síntomas incluyen, entre otros, confusión repentina, pérdida de habilidades, dificultad para comunicarse, debilidad en un lado o caída de la cara), busque atención médica de inmediato. Si cree que su estudiante ha tenido un derrame cerebral (recientemente o en el pasado), discútalo de inmediato con sus padres.

HCU NETWORK AMERICA

To download the full guide, visit: bit.ly/3EN3srf

NEW RESOURCE

Educator's Guide - Classical HCU

¡Ahora disponible en español!

Guía de HCU

Maestros

¿Qué es HCU?

La homocistinuria (oh-mo-cis-ti-nu-ria), o HCU, es una enfermedad genética metabólica rara la cual es hereditaria. Las personas con HCU no pueden descomponer los aminoácidos metionina (me-tio-ni-na) y homocisteína (oh-mo-cis-te-i-na) en sus cuerpos. La metionina (Met) se encuentra en la mayoría de los alimentos que contienen proteínas. HCU es una condición médica grave que se puede tratar con una fórmula médica especial para pacientes con HCU, una dieta baja en proteínas y Met, algunas vitaminas y otros medicamentos.

¿Qué sucede?

Normalmente, la metionina es descompuesta a otro aminoácido, la homocisteína (HCY) (oh-mo-cis-te-i-na). El subproducto homocisteína (HCY) también se acumula y tiene efectos secundarios peligrosos y dañinos cuando se ingiere proteína (específicamente Met). Los niveles altos de HCY pueden causar daños a los ojos, al sistema nervioso central, esquelético y al sistema vascula.

Los niveles altos de HCY pueden causar:

- Miopía severa (visión borrosa)
- Torpeza
- Dislocación del cristalino del ojo
- Extremidades largas
- Deficiencias cognitivas
- Coágulos de sangre
- Problemas de comportamiento
- Apoplejía (ataque cerebral)

Para ayudar a prevenir estos problemas, las personas con HCU deben seguir una dieta especial baja en proteínas y tomarse su fórmula médica para HCU durante todo el día.

¿Cómo puedo ayudar?

Enseñe a su estudiante como lo haría con cualquier otra persona. Los niveles de HCY pueden fluctuar. Su estudiante con HCU puede necesitar más tiempo o atención para seguir el ritmo de la clase.

Ayude a garantizar que su estudiante beba su fórmula médica. La presión social puede hacer que los niños tiren su fórmula en secreto o la oculten.

Es importante que su estudiante tenga un lugar donde se sienta seguro para guardar y consumir su fórmula.

Comuníquese con los padres y haga preguntas. Ya que usted como maestro pasa una buena cantidad de tiempo con su estudiante, es posible que sea el primero en notar problemas relacionados con la HCU. El manejo adecuado de la HCU depende de una colaboración entre padres y el personal de la escuela y que estos se comuniquen entre sí.

Celebraciones de la clase

Aunque su estudiante con HCU no puede comer galletas o pasteles comprados en la tienda, existen versiones bajas en proteínas de estos postres. Asegúrese de informar a los padres y al personal de la cafetería sobre una próxima celebración para que puedan proporcionar una merienda alternativa. Es posible que desee guardar unos dulces que no se echen a perder en el salón de clases para celebraciones inesperadas.

La fórmula HCU es una parte esencial de la dieta.

Dado que las personas con HCU no pueden tener muchos alimentos que contengan proteínas enteras, confían en la fórmula de HCU para suministrar proteínas libres de Met. La fórmula HCU generalmente se consume varias veces al día.

Consejos útiles

- ☐ Anime a su estudiante seguir su dieta. "Solo un bocado" no es recomendable para una dieta baja en proteínas. Infórmeles a los padres si su hijo no se come los alimentos enviados desde casa o si come algo que no fue enviado desde casa o acordado de antemano.
- ☐ Trátelos igual que a sus otros estudiantes. Su estudiante no está enfermo y no debe ser tratado como tal. Si sigue la dieta, puede tener tanto éxito como sus compañeros de clase.
- ☐ No permita que HCU defina a su estudiante. Establecer un sentido de uno mismo fuera de la HCU es una parte crucial de la autoaceptación.

HCU NETWORK AMERICA

To download the full guide, visit: bit.ly/3VA1LEV

INDUSTRY NEWS

Synlogic Announces SYNB1353 Achieves Proof of Mechanism for Treatment of Homocystinuria and Provides Business Update

Top-line Phase 1 data in healthy volunteers show that SYNB1353 reduces plasma methionine by consuming methionine in the GI tract

SYNB1353 has been granted Orphan Drug Designation (ODD) from the FDA for the treatment of homocystinuria (HCU)

CAMBRIDGE, Mass., Nov. 30, 2022 (GLOBE NEWSWIRE) -- Synlogic, Inc. (Nasdaq: SYBX), a clinical-stage biotechnology company developing medicines for metabolic and immunological diseases through its proprietary approach to synthetic biology, today announced that SYNB1353 has achieved proof of mechanism and positive results in a Phase 1 study in healthy volunteers treated with multiple ascending doses of SYNB1353. SYNB1353 is an orally administered, non-systemically absorbed drug candidate designed to consume methionine in the GI tract for the potential treatment of homocystinuria (HCU). The Company also shared that the FDA has granted Orphan Drug Designation (ODD) to SYNB1353 for the treatment of HCU. ODD is granted by the FDA to drugs or biologics intended to treat a rare disease or condition, which generally affects less than 200,000 individuals in the U.S. ODD-granted therapies entitle companies to development incentives including tax credits for qualified clinical trials, user fee exemptions, and the potential for seven years of market exclusivity after approval.

To read the full press release, visit: bit.ly/3FfEd24

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.

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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

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