

The HCU *Herald*

Featuring...



HCU Hero
Avery from Indiana



March 2026



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

HCU HERO: AVERY FROM INDIANA



My name is Avery. I am 26 years old and was born in April 1999 in Indiana. I was diagnosed with classical homocystinuria (HCU) in 2025. I do not believe HCU was included on the newborn screening panel in Indiana at the time I was born. Looking back, I had several symptoms consistent with classical HCU that were overlooked for years.

I am a PhD student at Cincinnati Children's Hospital Medical Center (CCHMC), where I study neurofibromatosis type 1. Working at the hospital allows me to see what clinical trials are ongoing. At the end of 2024, I learned about a clinical trial, CincyKidsSeq, that

offered free whole-genome sequencing. As both a student and a researcher, I was curious to learn more about my own genetics and wanted to contribute to science by participating.

In December 2024, I enrolled in the study and had my blood drawn for genomic sequencing. In 2025, while I was at work in the lab, I received a call informing me that my results were back and that the study wanted to schedule an appointment for me with a genetic counselor. My partner had participated in the study and received negative results directly through MyChart without a genetic counseling appointment, so I immediately suspected that I had some mutation, although I was not told what mutation I had at the time.

HCU HERO: AVERY FROM INDIANA

Before my scheduled appointment, I received another call explaining that I had two mutations in the CBS gene, meaning I possibly had homocystinuria. However, it was unclear whether both mutations were inherited from one parent or one mutation from each parent. Initially, my parents were asked to undergo genetic testing, but



because they live three hours away, this would have been difficult. Instead, the team decided to first test my homocysteine levels.

In early August 2025, I had my blood drawn, and just a few hours later my results came back showing my homocysteine levels were greater than 120 $\mu\text{mol/L}$. I remember seeing the notification while at work. It didn't feel real. After that, there were many follow-up phone calls and changes in the doctors I would be seeing. I am incredibly grateful to my boss, Elliott, who helped connect me with the appropriate specialists at CCHMC during this overwhelming time.

At my first genetics appointment, my doctor decided to begin a vitamin B6 responsiveness trial. I tested several doses, with blood draws every few days to monitor my response. To make the process less stressful, I had my family and friends guess what my homocysteine levels would be before each draw.

HCU HERO: AVERY FROM INDIANA

My baseline levels before the trial were again over 120 $\mu\text{mol/L}$. After a few days on 100 mg of B6, my levels dropped to 42.2 $\mu\text{mol/L}$. At 200 mg, they dropped to 37.3 $\mu\text{mol/L}$, and at 400 mg, they dropped further to 24.3 $\mu\text{mol/L}$. After the short trial, the doctor decided to have me try 200 mg for a longer period. After one and two months of 200 mg of B6, my homocysteine levels remained stable around 25 $\mu\text{mol/L}$. I am incredibly grateful to be a B6 responder.



Since my diagnosis, I was referred to an ophthalmologist. I have worn glasses since fifth grade, with progressively worsening vision, but thankfully I do not have lens dislocation. Prior to my HCU diagnosis, I had also learned that I have several spinal abnormalities, including scoliosis, a unilateral right pars defect at L5, and spina

bifida occulta, which was identified after months of back pain, physical therapy, and imaging. I also have long fingers and toes and have been diagnosed with anxiety and ADHD. While I exhibit many symptoms associated with HCU, I may have gone many more years without a diagnosis if I had not enrolled in this clinical trial and undergone genomic sequencing.

For many years, I have followed a mostly vegetarian and vegan diet because my body does not tolerate meat or dairy well, and I have continued this diet since my diagnosis.

HCU HERO: AVERY FROM INDIANA

Each morning, I now take vitamin B6, folic acid, and vitamin B12, along with my other medications. Fortunately, the transition has not been difficult. Although receiving an HCU diagnosis was frightening, I am deeply grateful for the support of my family, friends, and coworkers.



I now understand the increased risks associated with HCU, including blood clots, strokes, and pulmonary embolisms, and I am vigilant about monitoring symptoms. Despite everything, I have a more positive outlook on life. Learning that I have a genetic disorder, one even rarer than the condition I study, was scary, but the diagnosis was ultimately a blessing in disguise. Without this genetic testing, I likely would not have been diagnosed

until experiencing a serious, possibly life-threatening complication. By sharing my story, I hope to highlight the importance of genetic testing and help raise awareness about homocystinuria



FOOTBALL SQUARE FUNDRAISER



\$200

Q1 WINNERS: Holly Fulginiti

Q2 WINNERS: Sue Geyer

Q3 WINNERS: June Anderson

BOARD 1

\$400

GAME WINNERS: Mary Zelasko

\$100

Q1 WINNERS: Danae Bartke

Q2 WINNERS: Amy Nevitt

Q3 WINNERS: HCU Donation

BOARD 2

\$200

GAME WINNERS: June Anderson

Rare Disease Week

on

Capitol Hill



Rare Disease Week

on

Capitol Hill



Rare Disease Week

on

Capitol Hill



Are you ready to be the next young adult leader?



- Meet other young adults passionate about rare disease.



- Learn about roles and opportunities for patient representation in drug development, policy making, and the regulatory process.

- Engage with guest speakers in a small cohort. Become a more effective and powerful advocate.



**Classes meet Thursdays 6:00
-8:00 p.m. ET from April 23 - June
4, 2026**

**Open to
Advocates
Between 18
and 30 Years
of Age**



Interested in YARR year-round? Learn more and apply today! HearUsYARR.org

Apply Today!

YARR
Leadership Academy

ADVOCACY VIRTUAL WEBINAR SERIES

Tuesday, March 3rd
6:00 pm EST

REGISTER NOW

Advocacy Webinar Series – Collaboration in 2026

Workshop 1: Intro to Advocacy

Advocacy 101: What Advocacy Is—and How You Can Get Involved

Tuesday, March 3, 2026 - 6:00 PM EST
Live virtual webinar (Zoom)



Session Overview

Join HCU Network America, Propionic Acidemia Foundation, and the MSUD Family Support Group, in collaboration with the EveryLife Foundation, for a clear, approachable overview of advocacy. We'll cover the basics, common advocacy pathways, and simple actions you can take to support your community and drive change.

You'll learn

- What advocacy is (and what it isn't)
- Common ways advocates make an impact
- Easy first steps you can take right away
- Practical examples for rare disease communities

Register:

<https://us06web.zoom.us/join/register/FVKQHTWnR-umU9bh9qp3UQ>

Presented by:



PARTNERS



In collaboration with:



Advocacy Webinar Series – Collaboration in 2026

Workshop 2: Storytelling

Advocacy 101: Tell Your Story, Drive Change

Thursday, March 19, 2026 - 6:00 PM EST
Live virtual webinar (Zoom)



Session Overview

Your story is a powerful advocacy tool. Join HCU Network America, Propionic Acidemia Foundation, and the MSUD Family Support Group, in collaboration with the EveryLife Foundation, to learn simple storytelling frameworks, key message elements, tips for sharing your story with policymakers and others with confidence, and practice with your fellow advocates.

You'll learn

- How to shape your story for impact
- Key message elements that "stick"
- Tips for sharing with policymakers and partners
- How to communicate with confidence

Register:

<https://us06web.zoom.us/join/register/zkED4UJFQBiklyF5m7hyqQ>

Presented by:



PARTNERS

In collaboration with:



Thursday, March 19th
6:00 pm EST

REGISTER NOW

SCHOLARSHIP OPPORTUNITY



The Guthrie-Koch Scholarship



- Do you have Classical HCU?
- Are you a *high-school senior* or *current student* pursuing an undergraduate degree or technical school?

If you answered 'yes', you are eligible to apply for the Guthrie-Koch Scholarship Program!

The Guthrie-Koch Scholarship Program was founded in 1997 to recognize outstanding young adults with PKU pursuing higher education and provide financial support to these efforts, but has now been expanded to include young adults with Classical HCU and other metabolic disorders!

Click [here](#) to learn more and to start your application!

The application deadline is March 15, 2025.

[Click Here](#)

It's A

FAMILY AFFAIR

In The City of Brotherly Love



July 10-12, 2026
Philadelphia, PA

- Dedicated Science Day for Families and Professionals
- Scientific Poster Session
- HCU Hero Award Banquet
- Networking Opportunities
- Community fun day

- Patient Focused Panels
- Breakout Sessions
- KidsZone and Teen Zone
- Community Building Opportunities

Patient & Family Conference

REGISTER NOW



HCU
NETWORK
AMERICA

Classical HCU | Cobalamin Disorders | Severe MTHFR



HCU FAMILY Fun Day

On **Sunday, July 12**, to wrap up our biennial **HCU Network America 2026 conference**, we are hosting a fun way for our community to bond, laugh, and spend time together away from the hotel!

Join us for **activities and lunch** at Franklin Square from **10 am to 2 pm**.

We will be providing a continuous shuttle to and from the hotel for anyone who needs to return for departure.

If you would like to join us for this unforgettable experience, **please plan your travel accordingly.**

Carousel | Mini Golf | Yard Games

REGISTER NOW





**CALL FOR ABSTRACTS
SUBMISSION DEADLINE
APRIL 30th, 2026**

July 10-12, 2026



Abstract Submission Link

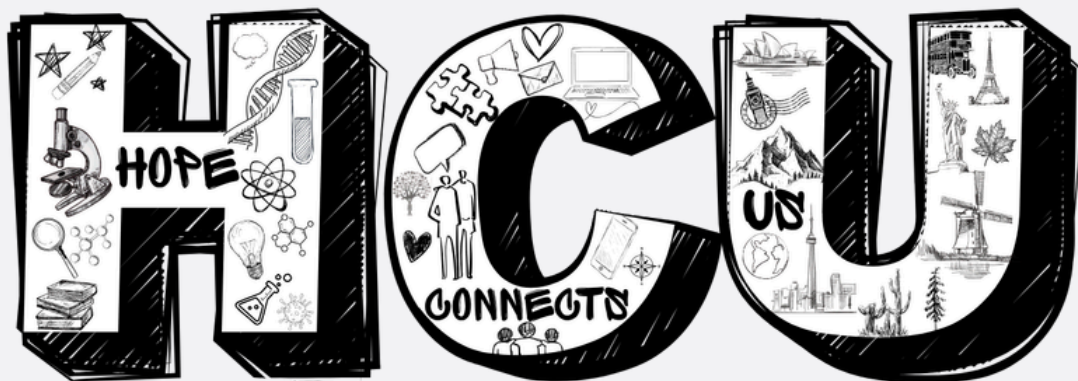
**For questions, email
research@hcunetworkamerica.org**

Posters on:

- **Natural history**
- **Novel therapies**
- **Treatment**
- **Diagnostics & Testing**
- **Other**

**Please note: accepted abstract
authors will need to register as a
medical professional or industry**

Hope Connects Us Around the World





World

Homocystinurias

Awareness Day

May 18th

-  **Livestream Event**
-  **Exclusive Raffle Baskets**

- ➔ Follow us to receive updated on activities & events
- ➔ Stay tuned for livestream details & raffle information

Together, *hope connects* us around the world.



Now Enrolling



Company: TraverreTherapeutics

Total time in study: About 9 months

Goal: To learn more about classical homocystinuria (HCU), and how safe and effective pegtibatinate may be in patients with HCU

Phase: This is a Phase 3 study that follows a Phase 1/Phase 2 study called COMPOSE

Who can take part?

- ✓ People with a diagnosis of HCU
- ✓ People 12 to 65 years of age

Other factors and restrictions also apply

What is the study drug?

- ✓ Participants will receive pegtibatinate or placebo (an inactive medication)

Why participate in the study?

- ✓ You may help others with HCU
- ✓ You will help contribute to new research and further understanding of HCU
- ✓ You will receive additional monitoring and testing



Study locations include: United States, Europe, the Middle East, Asia Pacific, and South America.



About 70 people with HCU will take part in the study.



Payment for time and travel may be available to participants and their caregivers.

Periods during the study



SCREENING PERIOD

To check if the right study is for you



PRE-TREATMENT PERIOD

To understand your current diet and how to keep it stable during the study



STUDY TREATMENT PERIOD

To receive pegtibatinate or the placebo



FOLLOW-UP PERIOD

To check your health after the Study Treatment Period

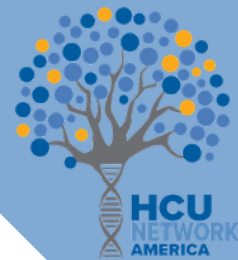
After completing the **HARMONY Study**, it may be possible for participants to join a long-term follow-on study and continue or start receiving pegtibatinate.

To find out more, please contact:
MedInfo@travere.com



For more information, please scan the QR code or visit:

www.hcuconnection.com



HCU NETWORK AMERICA PRESENTS CLINICAL TRIALS 101

WITH SAGAR VAIYDA, M.D. PH.D



MARCH 25



- **6 PM EDT**
- **11 PM GMT**
- **8 AM AEST**
- **(MARCH 26)**



REGISTER NOW >



HCU NETWORK AMERICA PRESENTS

HARMONY GLOBAL CLINICAL TRIAL

LIVE Q&A

WITH DR. JOSHUA BAKER
LURIE CHILDREN'S HOSPITAL



APRIL 17



- 4 PM EDT
- 9 PM GMT
- 6 AM AEST
- (APRIL 18)



REGISTER NOW >





HCU NETWORK AMERICA PRESENTS CLINICAL TRIALS IN MOTION

WITH SAGAR VAIYDA, M.D. PH.D AND BEN LEWIS



APRIL 29

REGISTER NOW >



- 7 PM EDT
- 12 AM GMT
- 9 AM AEST (APRIL 30)

WE WILL BE AT **ACMG 2026**

BOOTH: 1611

POSTER: P453

**“DIAGNOSTIC INTERVALS FOR PATIENTS WITH
HOMOCYSTINURIA: INSIGHTS FROM A
PATIENT-REPORTED DATA COLLECTION
PROGRAM”**

2026 ACMG Annual Clinical Genetics Meeting

Where Genetics Meets Innovation

March 10-14 - Baltimore





HCU Network America is pleased to announce they are

Now Accepting Request for Proposals

- Seeking novel proposals for treatment and prevention of clinical manifestations for **Classic Homocystinuria** and/or **Methionine Synthase deficiency** by exploring novel mechanisms to obtain proof of concept to enable progression to clinical trials



Application Deadline: May 31, 2026
2026 HCU Network America Grant
Application - Fill out form



MARCH MADNESS FUNDRAISER



March Madness Selection *Sunday 2026* is **March 15!**
Start assembling your pool & raise funding for resources,
education & outreach for **HCU Network America!**

March Madness is a three week period packed full of buzzer beaters and is a sports poolers dream. But how can you keep the excitement alive when most of your members have had their brackets busted? A great option is our Madness Squares pool format, as every game of the tournament will have a winner!



How Do Madness Squares Work?

If you are familiar with Super Bowl Squares, the main idea is the same for March Madness. A 10x10 grid of boxes is setup and each row and column is given a number from 0 to 9. Just like in Super Bowl Squares, each square of the grid can be claimed by a pool member.

Winner breakdown

Each round is worth a set number of points. You can determine this on your own, but be sure to let all of the entries know before the tournament begins what the scoring system will be. (You should write the point values under each round at the top of the bracket).

Declaring a Winner

Multiply the total number of correctly picked games in each round by the points assigned to that particular round. Tally all rounds together and the person with the highest point total wins!

- For further instructions and to print your bracket, visit:
<https://www.printyourbrackets.com/howtomarchmadness.html>
- For online tools, check out:
<https://www.runyourpool.com/march-madness-squares-pools.cfm>

IN CASE 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



Ben welcomes Amy to the table!

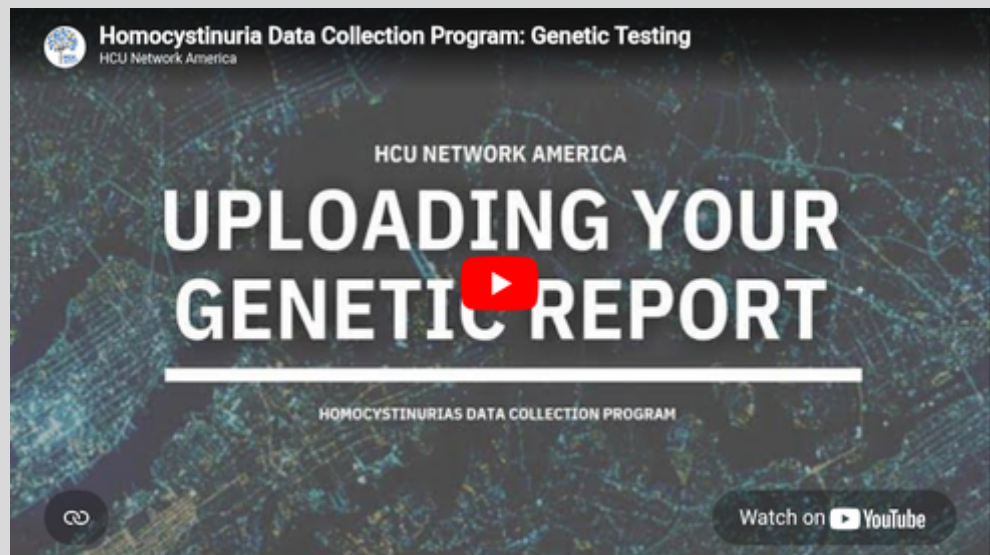
In this powerful and eye-opening episode, Ben sits down with Amy, the mother of Carson, to share their family's extraordinary diagnostic journey. For most of Carson's life, his symptoms were believed to be the result of Marfan Syndrome. It wasn't until genetic testing later on that the real answer emerged: Classical Homocystinuria (HCU).

UPLOADING YOUR GENETIC REPORT

Did you know a genetics report can help build a better understanding of homocystinuria?

Watch to find out:

- What is genetic testing?
- The components of a genetic report
- How I upload it into Rare-X
- Benefits of a genetic report
- Ways to get tested
- What you should expect



Upload your genetics report:
<https://rare-x.org/homocystinuria/>



Questions About RareX?



March 21, 2026 2:00pm-3:00pm MST

**Join our virtual open office hours
Our Research Coordinator is here to help!**



Brittany Parke
Research
Coordinator

**Join whenever
you can
during the
timeframe to
ask questions
or learn how
to navigate
RareX.**



**[Zoom Link](#)
[Click Here](#)**



PLAN YOUR PLATE

[CLICK HERE](#)

MONDAY

- CFL Penne a la Vodka Sauce (1.7 g protein)
- Promin Elbow Pasta (0.2 g protein)
- Capello's Gluten Free Focaccia Bread (< 0.5 g protein)
- Green Beans (0.9 per 50 g raw)

TUESDAY

- Zucchini, Mozzarella & Sun-dried Tomato Panini (3.33 g per sandwich)
- Crispy Yucca Fries (1/6 recipe equals 3.17 g protein without sauce)

WEDNESDAY

- CFL Minestrone Soup (2.5 g protein)
- CFL Cesar Salad (1.1 g protein)

THURSDAY

- Mushroom and Chive Risotto Swirled with Garlic Herb Butter

FRIDAY

- Schar Pizza Crust (1.51 g per 1/8 pizza base)
- Prego Pizzeria Style Pizza Sauce (1 g per 1/4 cup)
- Follow Your Heart, Dairy Free Mozzarella Style Shreds (0.6 g per 4 oz.)
- Add toppings to your liking - count as needed

- ### SNACKS
- Snyder Gluten Free Pretzels
 - Garden Veggie Straws
 - Siete Tortilla Chips
 - Apple Slices
 - Mandarin Orange

Notes

- All items and recipes are hyperlinked.
- All breakfast are 2 g or less
- All snacks are 1 g or less
- **Meals should be used for lunches and dinner to reduce time spent cooking.**
- Please verify all information using the flok app.

- ### BREAKFAST
- So Delicious Yogurt w/ Berries
 - Cambrooke Blueberry Scones
 - CFL Honey Puffs Cereal
 - CFL Porridge
 - Cambrooke Hot Cereal

RECIPE FEATURE

CLICK HERE

ZUCCHINI, MOZZARELLA & SUN-DRIED TOMATO PANINI WITH BASIL SAUCE

INGREDIENTS



2 tsp Italian Seasoning



2 Zucchini



4 Capello's Gluten Free Focaccia Bread



1 oz Basil



3 oz Sun-Dried Tomatoes in oil



2 tsp Garlic Powder



8 TBSP Follow Your Heart Mayonnaise



4 TBSP Follow Your Heart Sour Cheese



1 Cup Follow Your Heart Shredded Mozzarella

4 servings

- **Protein:** Zucchini, Mozzarella & Sun-dried Tomato Panini, 3.33 g protein per sandwich
- **Protein:** Crispy Yuca Fries, 1/6 recipe equals 3.17 g protein without sauce



Adapted from Hello Fresh

<https://www.hellofresh.com/recipes/zucchini-mozz-sun-dried-tomato-panini-5ee128214691fc476b4a9d30>

7/1/20 15:36

LOW-PROTEIN MUST-HAVES



What the focaccia? A flatbread that's this light, airy, and savory? Yet it's totally, incredibly gluten-free? How'd we pull off that magic trick? By using a special European wheat without gluten, that's how. Of course, we also added olive oil, rosemary, and flaky sea salt. So tear into this focaccia with wild abandon and say "Fuhrgheddelaboudit."

Cooking Instructions:
 1. Unwrap and thaw focaccia for 15 min while oven preheats to 375°.
 2. Place focaccia on baking sheet in middle rack of oven and bake at 375°.
 3. Bake for 18-24 minutes while enjoying the lovely aromatherapy.
 4. Serve immediately. Please try to refrain from talking with your mouth full.

Nutrition Facts
 About 7 servings per container
Serving size 1 oz (28g)
Amount per serving
Calories 150

% Daily Value*		% Daily Value*	
Total Fat 15g	30%	Total Carbohydrate 35g	70%
Sodium 150mg	30%	Dietary Fiber 1g	2%
Total Sugar 1g	2%	Total Sugar 1g	2%
Protein 1g	2%	Total Sugar 1g	2%

Ingredients: Whole Grain Free Wheat Flour*, Corn Starch, Olive Oil, Sea Salt, Yeast, Tapioca Flour, Sugar, Baking Powder, Eggs, Rice Bran Oil, Sunflower Oil, Soy Lecithin, Guar Gum, Rosemary, Garlic Powder, Contains Wheat.

KEEP FROZEN

Find



Nutrition Facts
 About 7 servings per container
Serving size 1 oz (28g)
Amount per serving
Calories 80

% Daily Value*		% Daily Value*	
Total Fat 7g	14%	Total Carbohydrate 5g	10%
Sodium 290mg	58%	Dietary Fiber 0g	0%
Saturated Fat 6g	12%	Total Sugars 0g	0%
Trans Fat 0g	0%	Includes 0g Added Sugars	0%
Cholesterol 0mg	0%	Protein 0g	0%

Ingredients: Water, Coconut Oil, Tapioca Flour, Sugar, Baking Powder, Yeast, Rice Bran Oil, Sunflower Oil, Soy Lecithin, Guar Gum, Rosemary, Garlic Powder, Contains Wheat.

Find

Nutrition Facts
 15 servings per container
Serving size 2.00
Amount per serving
Calories 90

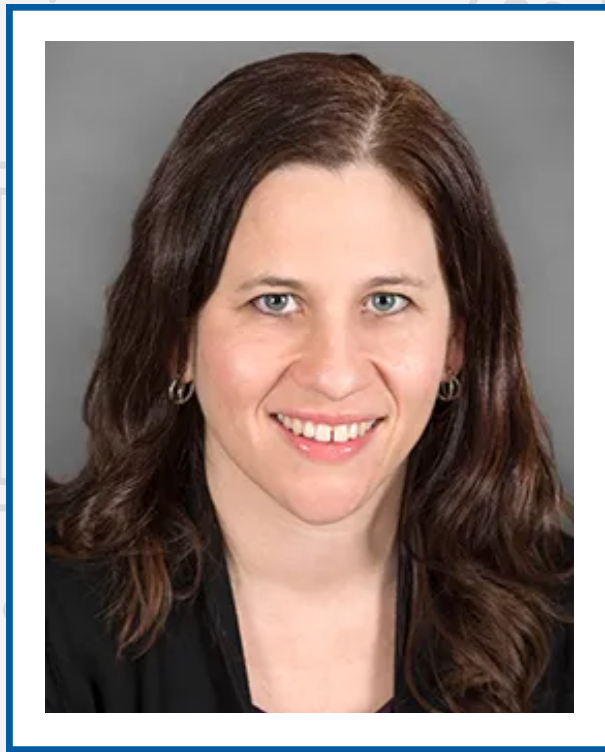
% Daily Value*	
Total Fat 9g	18%
Saturated Fat 4g	8%
Trans Fat 0g	0%
Cholesterol 0mg	0%
Sodium 25mg	0.5%
Total Carbohydrate 2g	4%
Dietary Fiber 0g	0%
Total Sugars 0g	0%
Includes 0g Added Sugars	0%
Protein 0g	0%

*The % Daily Value (DV) tells you how much a nutrient in a serving of food contributes to a daily diet. 2,000 calories a day is used for general nutrition advice.

Find



WELCOME OUR NEW *Medical Advisor*



Click Here

STEPHANIE SACHAROW

Dr. Stephanie Sacharow is a medical biochemical geneticist specializing in metabolic diseases, including phenylketonuria (PKU) and homocystinuria. She serves as Medical Director of the PAL Clinic at Boston Children's Hospital and directs the Dr. Harvey Levy Program for Phenylketonuria and Related Conditions.

Board-certified in Medical Genetics, Medical Biochemical Genetics, and Pediatrics, Dr. Sacharow has led newborn screening initiatives, published extensively, and serves as a co-investigator on multiple clinical trials. She is a recognized expert in pegvaliase (Palynziq) management and a co-author of treatment guidelines, frequently speaking at national and international conferences.



PUBERTY TO PREGNANCY TO MENOPAUSE

FOR INHERITED METABOLIC DISORDERS

18 YEARS OR OLDER

APPROXIMATELY 10-15 MINUTES

HELPS ADVANCE REPRODUCTIVE HEALTHCARE



Click Here

TAKE ACTION: IMPORTANT POLICY

#Foods&FormulasAccess

#MedicalNutrition



**RAISE
YOUR VOICE**



&

Take Action

Support the
**MEDICAL FOODS AND
FORMULAS ACCESS ACT**

HR 5684 / S 3304

CLICK HERE

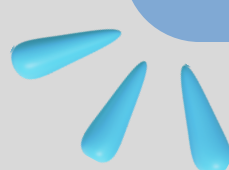


IMPORTANT ANNOUNCEMENT

Congress passed legislation that includes a five-year reauthorization of the Rare Pediatric Disease Priority Review Voucher (PRV) Program!

Congress has passed legislation that includes a five-year reauthorization of the Rare Pediatric Disease Priority Review Voucher (PRV) Program, increased funding for rare disease research, and key protections for NIH funding.

The PRV Program has helped advance treatments for 40+ rare pediatric diseases at no cost to taxpayers, bringing hope to thousands of families. Its reauthorization means stalled research and clinical trials can now move forward.



CUSTOMIZE YOUR KIT FOR FREE!



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!*

- **What is it?**
 - A secure private survey for individuals or families affected by Homocystinuria
- **What will I share?**
 - Patient's birthdate, gender, exact diagnosis, and how they were diagnosed
- **What will my info be used for?**
 - Confidential and will not be shared unless we have permission
 - Helps HCUNA achieve our goals

- **Why should I join?**
 - Able to find other families and patients in your state and request contact information
 - Access to exclusive materials (ex: we may have a webinar that a presenter doesn't want to share publicly but is okay sharing with just our community)

What?

Why?

Contact Register

How?

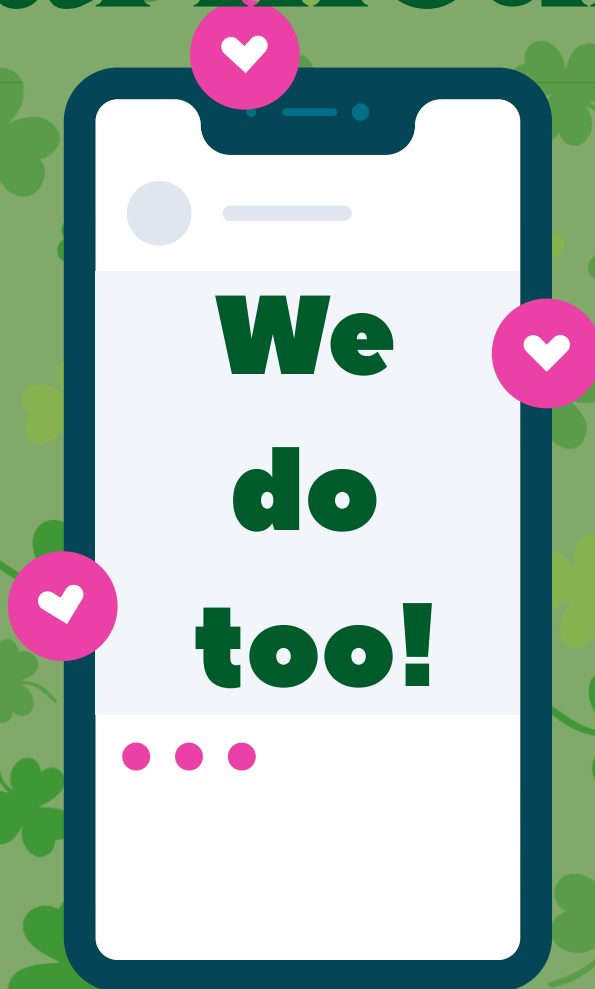
- **How do I participate?**
 - The form takes 3-5 minutes to complete
 - Visit our website and click on "contact register" tab or...

[Click Here](#)

Follow Us!



Do you have social media??



Follow us 



@hcu_network_america



HCU Network America



@HCUAmerica