The HCU Herald

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

HCU Network America

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HCU HERO:
JORDAN FROM KENTUCKY
DECEMBER 25, 1996- JUNE 24, 2020

Jordan came into this world on a snowy Christmas morning in 1996. He was the youngest of two children, ten years apart in age, and the youngest grandchild on both sides of the family. From the very beginning, Jordan was always the one to push the limits, march to the beat of his own drum and defy odds over and over again. As a newborn, Jordan passed all newborn screenings with flying colors but as he grew his family began to notice he was not meeting some of his developmental milestones. Jordan's pediatrician referred him for a developmental evaluation around three years of age. The result of this evaluation revealed global developmental delays, as well as significant vision impairment. Jordan had always been a happy baby and was very interactive with his environment. Vision impairment was never really something we had considered until he got his first pair of glasses at three years old; the light in his eyes to see the world around him told us how much he'd been missing.

After the official diagnosis of global development delays, Jordan began his journey to learn new skills whether that be running, jumping, social skills, balance, or language development. His favorite was hippotherapy as he sometimes got to ride horses backwards while catching balls midair! Genetic testing had also taken place after Jordan's initial diagnosis, but results yielded negative findings for common genetic disorders. At the age of six, he woke up one morning and noted that he couldn't see which sent him on an emergency visit to a pediatric ophthalmologist. At this time, his family learned that Jordan had displaced lenses in both eyes. This was a hallmark characteristic associated with something called Homocystinuria (HCU) and he was referred to Riley Children's Hospital in Indianapolis for further follow up care. Jordan underwent additional genetic testing for rare diseases and eye surgery in Indianapolis to remove both detached lenses. After surgery, Jordan was considered legally blind without glasses, but had some visibility with a special type of lens. Jordan always had 3 pairs of glasses: one primary, one secondary (in case the first pair broke as things do with small children) and sports glasses. These glasses were very costly and not always fully covered by insurance which often was the case for many of the therapeutic treatments Jordan received for HCU.

Riley Children's was able to confirm HCU as Jordan's official diagnosis at the age of six through bloodwork, urine and genetic testing. HCU had been missed in his pediatric screening because, at the time, HCU was not something routinely assessed in infants in the US. The prevalence of classic HCU is 1 in 200,000 people. Armed with this new information, Jordan and his family finally had a diagnosis and a treatment path needed to help Jordan be successful and healthy. Having classic HCU meant his disorder had to be controlled through a low protein diet, B6, B12, folic acid, Cystadane and Hominex formula to supplement his nutritional needs. Like so many others, Jordan disliked the taste of the formula, but powered through it for many years. He would drink the formula in various mixed drinks but finally resorted to vegan capsules/pill form. To continue receiving the correct amount of formula and to keep his blood levels stable he would take 120 pills a day. At the time, formula mostly came in powdered form and pharmacies were not at liberty to stuff capsules although some smaller family-owned pharmacies were willing to help us out from time-to-time. As a family, we all took turns filling each capsule with

white formula powder by hand; whatever it took we were going to get the job done! Jordan also used medical foods which are created in a laboratory for those who have supplemental nutritional needs. Although medical foods are created in a laboratory and are considered medically necessary for those that consume them, most insurances do not consider medical foods to be a prescription drug, therefore, eliminating coverage for many. In some instances, Jordan's parents were able to get reimbursed on the backend for these medical foods but often times not.

Through Jordan's own strong will, the support of family AND friends, and numerous specialized healthcare providers, Jordan was thriving. Jordan was able to graduate from high school in 2017. He loved bowling for countless hours every day and worked at a bowling alley; his best score being 225, but always working towards that 300! He played the guitar and aspired to be a country music star, traveling the road and playing songs at different venues. He liked school a lot and wanted to find a college that would be a good fit for him to explore his interests and meet new friends. He was a french fry connoisseur and always knew the best places for a good fry. Jordan loved to travel and would travel to Minnesota every year where he learned to fish like his mother, uncles, sister, and cousins. Jordan and his family took a Make-A-Wish trip out West in an RV to see several national parks. Traveling to the wild west was a dream of Jordan's as his favorite movie was The Shakiest Gun in the West, staring Don Knotts. Jordan watched all of Don Knott's movies and loved Mayberry RFD. He was an avid movie lover too, and you would be hard pressed to find a movie he hadn't seen! Other activities Jordan was involved in included: swimming, archery, song writing, WWE wrestling, collecting comic books, and following college and professional football/basketball. GO WILDCATS!

During the years 2017-2020, Jordan had begun to struggle with diet and formula compliance. He was burnt out on the diligence required and the taste of his formula which gave him acid reflux. Despite the efforts of his family and physicians, he would yo-yo with his diet and formula consistency. In January of 2020, Jordan began experiencing soreness in his back and limbs which physicians attributed to muscle overuse. In April of 2020, we noticed mysterious bruises all over his body and took him to the local emergency room where he was diagnosed with Acute Lymphoblastic Leukemia (ALL) t-cell. Rapid treatment began to take place to treat his cancer as ALL is very aggressive. Jordan started his cancer treatments for the first month in Louisville, KY but eventually transferred to Cincinnati Children's Hospital due to their expertise in ALL and HCU. Jordan is the first documented case in the world for having both HCU and ALL t-cell. Many chemotherapy options were contraindicated for HCU due to the counter effects that would put Jordan at risk for strokes and blood clots; this greatly hindered his care team's ability to manage both disorders effectively. Despite the world being locked down from COVID, on June 24th, 2020, Jordan passed away surrounded by his parents and sister.

Jordan leaves behind a once in a lifetime experience for both his family and friends. Knowing Jordan has inspired several family members and friends to go into healthcare. His treatment journey will hopefully shine a light on the path for those to follow him. Most importantly, is the continuous need for contributions to science furthering the equity of healthcare for those with HCU and the appreciation for individuals with HCU, as well as their families, as they are pioneers for this rare disease.

Email us your patient story! info@hcunetworkamerica.org

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: Muffin & Banana
Lunch: Grilled Cheese & Soup
Dinner: Baked Potato

T

Breakfast: "Eggs" & Strawberries
Lunch: Pasta Salad
Dinner: Tacos

W

Breakfast: Waffle & Fruit
Lunch: Avocado Toast
Dinner: Mac and "Cheese" with Broccoli

T

Breakfast: Cereal with Fruit
Lunch: Sandiwch & pretzels
Dinner: Asian Stir Fry

F

Breakfast: Eggz Sandwich
Lunch: Nuggets and veggies
Dinner: Make your own pizza

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.

Corn Salsa

***Yields 8.4 servings* | Serving Size: 1 oz. (28g) | Protein per serving: 0.5 g | Calories per serving: 23**

Ingredients:

- 1 c Canned Corn, rinsed
- 1 TBSP Diced Raw Onions
- 2 TBSP Diced Raw Bell Peppers
- 1 TBSP Diced Jalapeno
- 2 tsp Cilantro, chopped
- 1 TBSP Lime Juice
- 1/2 tsp Chili Powder
- 1/4 tsp Ground Cumin
- 1 tsp Apple Cider Vinegar
- 1/2 oz. Follow Your Heart Dairy Free Feta Crumbles

Directions:

1. Combine all ingredients except feta crumbles in a sealable glass bowl. Season with salt and pepper to taste. Refrigerate overnight to allow flavors to meld. Top with feta crumbles and serve chilled

Jackfruit Nuggets

Yields 1 serving | Serving size: 4 nuggets
Protein per serving: 1.1 g | Calories per serving: 169

Ingredients:

- 4 Nugget Jackfruit Chik'n Nuggets Mixture, as prepared and frozen (see below)
- 2 fl.oz. Rice Milk
- 2 TBSP Cook for Love Toasted Bread Crumbs (*https://cookforlove.org/recipes/detail/75564*)

Note: you can sub other bread or use already made bread crumbs, but will need to adjust the protein content.

Directions:

1. In a small skillet, heat vegetable oil over medium heat. You want at least 1/2 inch of oil to fry the nuggets.
2. Place the rice milk (or any non-dairy milk) in a small bowl and the breadcrumbs in another small bowl. Dip each nugget in the rice milk, then coat with the breadcrumbs. Place the nuggets in the preheated skillet. Fry until golden brown on both sides. Remove nuggets and place on a paper towel lined plate to absorb the grease. Serve warm with your choice of dipping sauce.

Jackfruit Chik'n Nuggets Mixture

Yields 21.4 servings
Serving size: 1 nugget
Protein per serving: 0.2 g
Calories per serving: 12

Ingredients:

- 1 20-oz. can Jackfruit, drained, seeds removed
- 1/3 c Cambrooke Chicken Patty Mix
- 2 tsp Cambrooke Chicken-Flavored Consomme & Seasoning, dry

Directions:

1. Rinse the jackfruit well and remove the seeds. Squeeze the excess liquid from the jackfruit and place in a food processor. Pulse the jackfruit until finely chopped, but not into a paste. Empty into a medium bowl. Add the consomme seasoning and chicken patty mix. Mix well with your hands or spatula.
2. Line a baking sheet with parchment paper. Using a scale, measure each nugget to be 1/2oz or 14 grams in weight. Shape them into rounds and flatten to resemble a chicken nugget. Lay on the parchment and repeat to the rest of the mixture. Freeze until ready to use.

UPCOMING EVENTS

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

Low Protein Cooking Class

July 16 | Time: 2 PM EDT

Fresh from the Farmer Market and Low Protein Grocery Store Finds

With Chef Amber

Cobalamin Disorders with HCU Community Virtual Meet-up

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

Saturday, July 30, 2022 | 8 am PT | 11 am ET | 4 pm UTC+1

Classical HCU Parent-Caregiver Meetup

Parents,Grandparents and Caregivers of "kids" all ages with HCU need support too!

August 20, 2022 at 10 am CT

Come network and learn from other parents, grandparents and caregivers!

Classical HCU Community Virtual Meet-up

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

Sunday, August 21, 2022 | 12 pm PT | 3 pm ET | 8 PM UTC

SEPTEMBER 1-30, 2022

More swag available for fundraisers!

Per Individual: $30

Per Family (up to 4 – 1 mailing address): $50

Registration now!

https://runsignup.com/Race/IL/Batavia/HCURaceforResearch

THE 2ND ANNUAL ROBERT GUTHRIE SYMPOSIUM

Hosted by JOHN R. OISHEI Children's Hospital | Jacobs School of Medicine and Biomedical Sciences

JUNE 28, 2022 | 8 A.M. – 5 P.M.

U.B. Jacobs School of Medicine and Biomedical Sciences
M&T Auditorium

JOIN US TO MEET THE EXPERTS AND EXPAND YOUR KNOWLEDGE

The 2022 Robert Guthrie Symposium is an all-day, hybrid educational event honoring the life and legacy of Robert Guthrie, MD, PhD, and his attachment to the University at Buffalo and Oishei Children's Hospital.

This annual symposium is a forum for world-renowned physicians and scientists to share their knowledge with providers, families and community stakeholders. Our speakers will discuss pioneering research and clinical practices in genetic and metabolic disorders.

This year, our focus is on the past, present, and future of newborn screening and metabolic disorders. The goal of the event is to educate professionals and patients on metabolic disorders and their treatment options, as well as, the history and current practices of newborn screening.

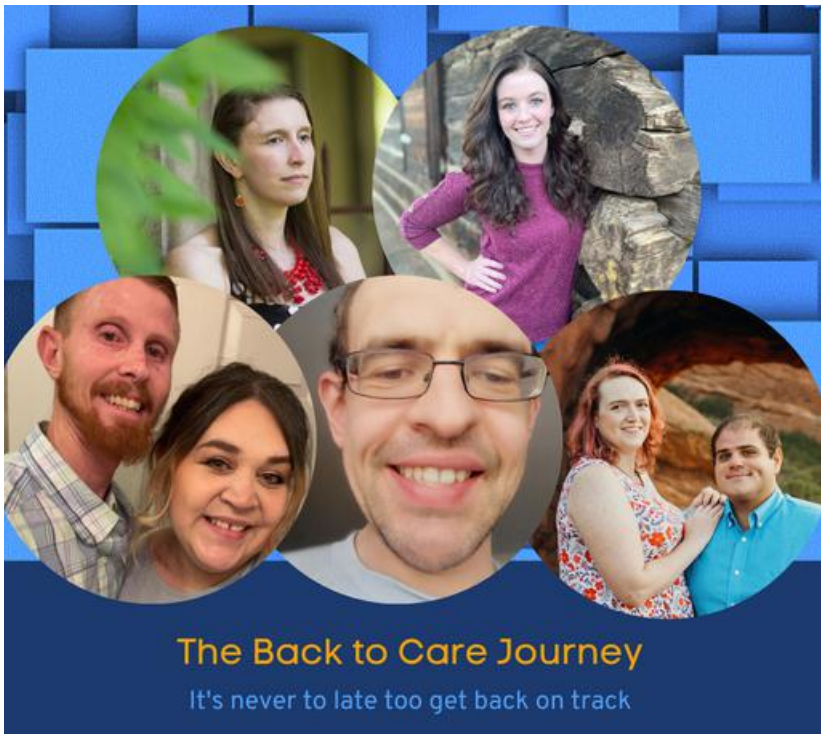
HCU Network America's Board President, Margie McGlynn, will chair the afternoon session for this symposium which will include presentations on new therapies for metabolic diseases. Margie's sisters were treated at Oishei Children's Hospital, who is sponsoring the symposium, before they passed away in the early 1970's

SIGN UP TODAY

Scan the QR code to register and for more information about the event.

https://www.ub-connect.com/s/1703/alumni/index.aspx?sid=1703&gid=2&pgid=3792&content_id=3813

EVENT RECAPS



Back to Care Panel Discussion

Back in April, Classical HCU patients Valerie, Janet, Ashley and Aimee joined HCU Network America's Executive Director and adult HCU patient, Danae' Bartke as they talked about their experiences surrounding Back to Care.

Patients talked about how the timing of their diagnosis impacted their issues with compliance, but even with access and a supportive family it was still a struggle. The one thing they all felt though was that there was hope for those who wanted to navigate back to care.

Watch the recording here:
https://youtu.be/C70zaoRY9Wc

Thank you for Going Blue for HCU!

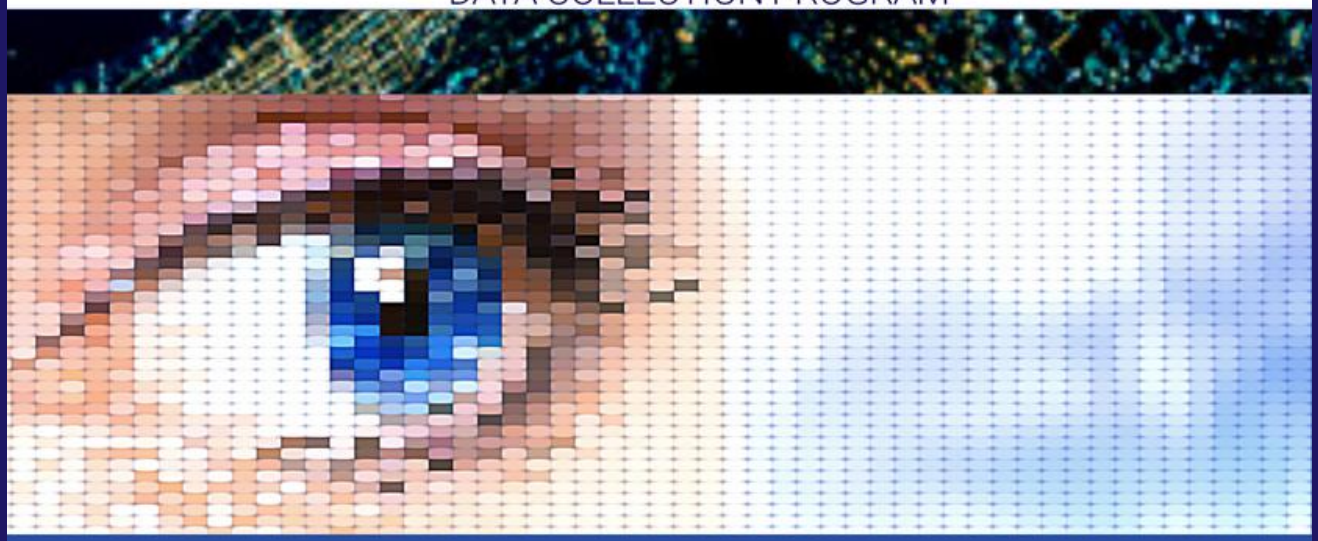
We'd like to thank everyone who participated in the 2022 Go Blue for HCU Campaign!

Between our t-shirt raffle, t-shirt sales, and peer to peer campaigns, we were able to raise over $1,000 for research!

There are still a few days left to get this limited edition Go Blue for HCU shirt! Get yours now https://www.bonfire.com/go-blue-for-hcu-2022/

RESEARCH OPPORTUNITIES

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

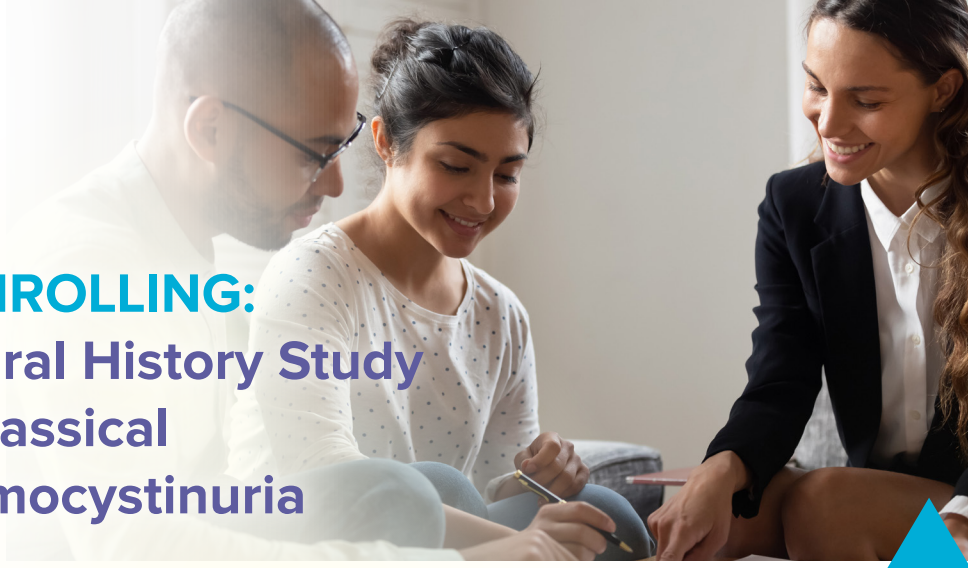


LET US SEE THINGS FROM YOUR PERSPECTIVE

From droopy eyelids (ptosis) to severe vision impairment, patients with various types of homocystinuria experience a wide range of eye issues. While some of these eye issues are better documented, other aspects aren't. We want to hear from you - help us see things from your perspective!

Complete the Eyes and Vision Survey
homocystinuria.rare-x.org

HCU Network America HCU Network Australia POWERED BY RARE-X



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

ICYMI RESEARCH NEWS

HCU Network America, OAA and CblC Onlus Announce First Collaborative CblC Research Grant

May 13, 2022 - CblC Onlus, HCU Network America and the Organic Acidemia Association announced the recipient of their first collaborative cobalamin research grant– awarding a research grant to the National Research Council's Institute of Biophysics in Palermo, Italy to identify potential treatment for cobalamin C (cblC) deficiency. The research, led by Dr. Silvia Vilasi, aims to identify compounds that could potentially rescue MMACHC functional deficiency in cblC disease. Dr. Vilasi is a researcher at the Institute of Biophysics (IBF) in the National Research Council, and has had a longstanding interest and involvement in the study of structure-function relationship of proteins involved in human pathologies. More recently she focused her interest in homocystinuria research.

Read the full press release at: https://hcunetworkamerica.org/wp-content/uploads/2022/05/2022-Cblc-Research-Grant-PR-Issued-5.13.2022-Final.pdf

Travere Therapeutics to Present Abstracts at the Society for Inherited Metabolic Disorders 43rd Annual Meeting and the Genetic Metabolic Dieticians International Conference 2022

April 6, 2022 - Travere Therapeutics, Inc. announced that the Company will present data from the ongoing Phase 1/2 COMPOSE Study of pegtibatinase, a novel investigational enzyme replacement therapy being evaluated for the treatment of classical homocystinuria (HCU), at the Society for Inherited Metabolic Disorders (SIMD) 43rd Annual Meeting, and the 2022 Genetic Metabolic Dieticians International (GMDI) Conference. In December 2021, the Company announced positive topline results from the COMPOSE Study. The Company and its collaborators will also present real-world evidence from metabolic centers of excellence on current challenges in the dietary management of classical HCU.

At GMDI the Company had two presentations titled:

- Pegtibatinase, an Investigational Enzyme Replacement Therapy for the Treatment of Classical Homocystinuria: Initial Results from the Phase 1/2 COMPOSE Study. Oral Session: Building Evidence for Evidence-Based Practice Clinical Research
- Dietary goals and current challenges in the management of classical homocystinuria: insights from multinational real-world experience

At SIMD the Company presented 1 poster titled:

- Pegtibatinase, an Investigational Enzyme Replacement Therapy for the Treatment of Classical Homocystinuria: Initial Results from the Phase 1/2 COMPOSE Study

To view the full press release visit: https://ir.travere.com/node/13481/pdf
Please email IR@travere.com to inquire about copies of posters.

Synlogic to Present Data on Phenylketonuria and Homocystinuria Programs at the Society for Inherited Metabolic Disorders 43rd Annual Meeting

April 1, 2022 - Synlogic, Inc., a clinical-stage biotechnology company developing medicines for metabolic and immunological diseases through its proprietary approach to synthetic biology, announced that data from its phenylketonuria (PKU) and homocystinuria (HCU) programs will be highlighted in two poster presentations at the Society for Inherited Metabolic Disorders (SIMD) 43rd Annual Meeting held April 10-13, 2022 in Orlando, Florida.

View the full press release here:
https://hcunetworkamerica.org/wp-content/uploads/2022/05/2022-Synlogic-SIMD-Press-Release-SYNB1353.pdf

70

Activity of SYNB1353, an Investigational Methionine-Consuming Synthetic Biotic Medicine, in an Acute Nonhuman Primate Model of Homocystinuria

Mylene Perreault[1], Erik Gerson[1], Jillian Means[1], Michael James[1], Ted Moore[2], Analise Reeves[1], David Lubkowicz[1], and David Hava[1]
[1]Synlogic Inc, Cambridge, MA, USA [2]Ginkgo Bioworks Boston, MA, USA.

Introduction

Homocystinuria (HCU) is a recessive inherited disorder caused by a defect in cystathionine β-synthase (CBS), which results in abnormal methionine metabolism and leads to an accumulation of homocysteine (Hcy) in the body (**Figure 1**). Elevated Hcy levels are associated with impairments of the eye, skeletal system, vascular system, and central nervous system. In patients with residual CBS activity (~50% of HCU population), vitamin B6 (pyridoxine) is effective at reducing Hcy levels. For pyridoxine unresponsive patients, betaine (involved in remethylation of Hcy to methionine) and a low-methionine diet that is very low in natural protein are the current therapeutic options. Early initiation of a low-methionine diet significantly lowers the risk of developing complications in HCU patients, but compliance to low protein diet is difficult.

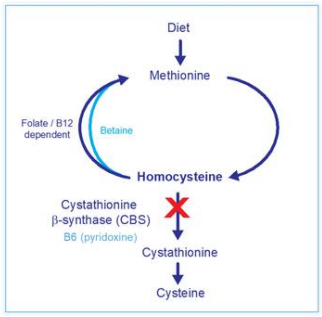


Figure 1. In HCU patients, mutations in the *CBS* gene result in accumulation of Hcy. Pharmacotherapeutic options for the treatment of HCU consist of vitamin B6 (pyridoxine), which can lower Hcy levels in B6-responsive patients, and betaine, which is involved in Hcy remethylation to methionine.

Study Design

The probiotic *E. coli* Nissle (EcN) was engineered to metabolize methionine within the gastrointestinal (GI) tract via the methionine decarboxylase (MetDC) pathway (**Figure 2**). Using proprietary codebase and metagenomic libraries, combined with protein engineering strategies, MetDC from *Streptomyces sp. 590* and methionine importer MetP from *Flavobacterium segetis* were identified by metagenomic screen and MetDC was further optimized via protein engineering. Genes encoding these proteins were chromosomally integrated under the control of a chemically inducible promoter, P_{tac}, which is induced by isopropyl β-D-1-thiogalactopyranoside (IPTG). To prevent the release of methionine in the GI tract once it enters the cell, the *yjeH* gene that encodes a methionine/branched chain amino acid exporter was deleted. The resulting strain, SYNB1353, converts methionine to carbon dioxide (CO_2) and 3-methylthiopropylamine (3-MTP), which is used as a biomarker of strain activity.

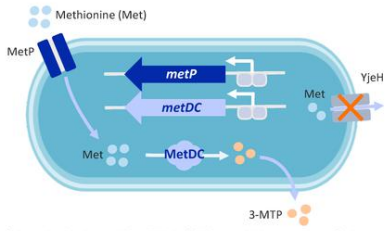


Figure 2. Schematic of engineered *E. coli* Nissle SYNB1353 with its components. Optimal metP and metDC were identified using proprietary metagenomic, codebase and protein engineering libraries.

Results

Engineered EcN SYNB1353 consumes methionine and produces 3-MTP *in vitro*

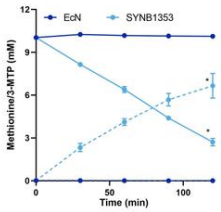


Figure 3. In vitro methionine consumption (solid line) and 3-MTP production (dotted line) by EcN (control bacteria) or SYNB1353. Cells were incubated for the indicated time with 10 mM methionine at 37°C, and supernatants were collected. *p<0.05 versus EcN.

An oral methionine load increases plasma methionine (A) and plasma total Hcy (B) in healthy cynomolgus monkeys

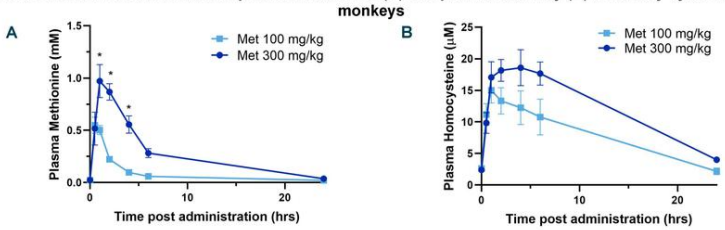


Figure 4. Monkeys were fasted overnight and received a single oral dose of methionine (100 or 300 mg/kg). Blood was collected for methionine and Hcy measurements. Data presented as mean ± SEM (n = 6/group). Statistical analysis was performed using two-way repeated ANOVA with Sidak's multiple comparison test. *$p < 0.05$ versus 100 mg/kg methionine.

SYNB1353 is active in a nonhuman primate model of acute homocystinuria

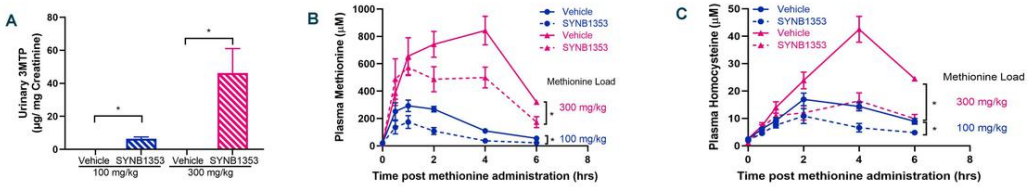


Figure 5. Monkeys were fasted overnight and received a single oral dose of methionine (100 or 300 mg/kg) with vehicle or SYNB1353 (1 x 10^{12} live cells). Urine was collected for 3-MTP, and blood was collected for methionine and Hcy measurements. Data presented as mean ± SEM (n=12/group for 100 mg/kg methionine, n=6/group for 300 mg/kg methionine). Statistical analysis was performed using unpaired t-test with Welch's correction (A) and two-way ANOVA with Sidak's multiple comparison test (B-C). *$p < 0.05$.

SYNB1353 dose-dependently increases urinary recovery of 3-MTP (A) and decreases plasma methionine (B) and plasma homocysteine (C) in a nonhuman primate model of acute homocystinuria

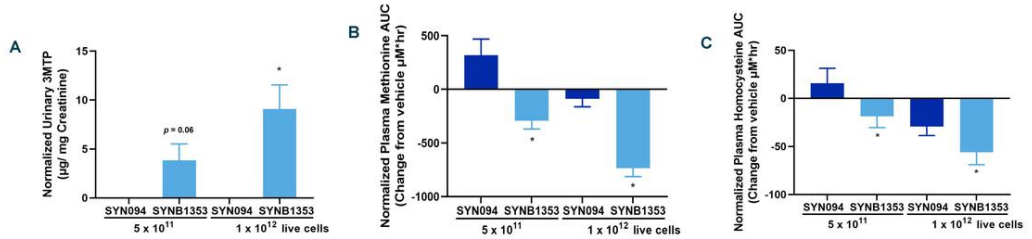


Figure 6. Monkeys were fasted overnight and received a single oral dose of methionine (100 mg/kg) with vehicle, EcN (control strain) or SYNB1353 (5 x 10^{11} or 1 x 10^{12} live cells). Urine was collected for 3-MTP, and blood was collected for methionine and Hcy measurements. Data was normalized to the study-respective vehicle and presented as mean SEM (n = 12/group). Statistical analysis was performed using paired t-test. *$p < 0.05$.

Conclusions

SYNB1353 is an engineered *E.coli* Nissle strain capable of metabolizing methionine and producing 3-MTP in vitro.

Concomitant administration of SYNB1353 with an oral load of methionine blunts the appearance of plasma methionine and plasma total homocysteine in nonhuman primates. Thus, SYNB1353 represents a promising approach for the treatment of HCU.

NOW AVAILABLE

Betaine Anhydrous for Oral Solution

*The **FIRST AB RATED GENERIC** Version of*
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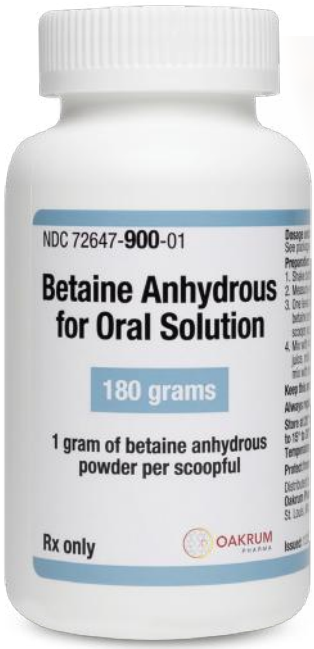

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72647-900-01	1 gm/scoopful	Powder	AB	CYSTADANE®

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INDICATIONS AND USAGE

Betaine Anhydrous for Oral Solution is indicated for the treatment of homocystinuria to decrease elevated homocysteine blood concentrations in pediatric and adult patients. Included within the category of homocystinuria are:

- Cystathionine beta-synthase (CBS) deficiency
- 5,10-methylenetetrahydrofolate reductase (MTHFR) deficiency
- Cobalamin cofactor metabolism (cbl) defect

IMPORTANT SAFETY INFORMATION

WARNINGS AND PRECAUTIONS

Hypermethioninemia in Patients with CBS Deficiency: Betaine Anhydrous for Oral Solution may worsen elevated plasma methionine concentrations and cerebral edema has been reported. Monitor plasma methionine concentrations in patients with CBS deficiency. Keep plasma methionine concentrations below 1,000 micromol/L through dietary modification and, if necessary, a reduction of Betaine dosage.

ADVERSE REACTIONS

Most common adverse reactions (> 2%) are: nausea and gastrointestinal distress, based on physician survey.

To report SUSPECTED ADVERSE REACTIONS, contact Oakrum Pharma, LLC at 1-833-444-8010 or FDA at 1-800-FDA-1088 or www.fda.gov/medwatch.

Click here for full prescribing information.

OAKRUM PHARMA

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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://hcunetworkamerica.org/contact-register/

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