The HCU Herald

Featuring...

HCU Heroes

Nicholas & Connor from Australia

June 2023

HCU NETWORK AMERICA

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

HCU HEROES:
NICHOLAS & CONNOR FROM AUSTRALIA

Our third son, Nicholas was born in the early hours of November 19th, 2019, in Sydney. The labour was straightforward, and we were sent home with our beautiful bundle later that day. My husband Daniel had some time off work during those 2 weeks, and we adjusted to now being a family of 5. Everything seemed to be going smoothly.

The day that Daniel returned to work, I received a call from the local hospital asking me to bring Nicholas back in the following day for another test as his original newborn screen had flagged an issue. We had a friend over later that day to meet our new baby and we discussed the call and our concern that something could be wrong. Our friend had a similar occurrence with their son and when they had the follow-up test, it turned out that it was a lab issue, and everything was perfectly fine.

The following morning we took Nicholas for the blood tests and the nurse suggested our baby may just have a B12 deficiency. We did the follow-up heel prick test, and we took him home feeling relieved that it was probably nothing serious, particularly as he seemed a happy baby.

Two days later I received another call; this time from a doctor at the children's hospital as well as someone from newborn screening, and they both seemed very concerned. They asked me if Nicholas was ok, if he was sick and feeding properly and wanted me to bring him in to the children's hospital urgently. When we arrived at the hospital we didn't wait long before we were ushered into a room filled with doctors, nurses and a social worker. At this moment we felt this was something really serious and were terrified! The doctors were amazed at how healthy Nicholas

seemed to be, but they quickly took him off for blood tests and to start injections of B12. My husband went with Nicholas and the doctors because I was an absolute blubbering mess and stayed behind with the social worker. I have no idea what we discussed because I think I was in shock. An hour later Daniel returned and they explained that they wanted to admit Nicholas,. so we would be staying at the hospital. We met with Dr. Ellaway, the head of the metabolic department. Dr. Ellaway explained that Nicholas had a metabolic disorder, and they weren't sure which one, but wanted us to stay in for tests to see if he would respond to the B12 injections.

The blood test results showed that Nicholas was b12 responsive, so this pointed to him having Cobalamin C disorder. We discussed the importance of his medication and they explained to us all the possible side effects and what to watch out for. We were discharged after 3 days and going home was scarier than it had ever been going home with a newborn. We had so many worries about his feeding (What if he got sick? What does all this mean for our new baby?). All of these scenarios were running through our minds.

Over December and into early January we got to know the team at Westmead Children's Hospital as we were regular visitors for checkups, blood tests, and injections. In late January 2020, we relocated from Sydney to Queensland – a move that had been planned since I first found out I was pregnant. We had committed to the move, but we were very worried about leaving Sydney and what this could mean for Nicholas' continuity of care.

...going home was scarier than it had ever been going home with a newborn. We had so many worries about his feeding (What if he got sick? What does all this mean for our new baby?). All of these scenarios were running through our minds.

After about a month of living in Brisbane we were contacted by the Queensland Children's Hospital and went for our first visit. We were still in the process of wrapping our heads around what this all meant, but I was really keen to have my

other two sons tested to make sure they were ok. The doctors explained to us that we shouldn't be worried about our other sons, that Nicholas was our main priority, and that they would go back and double-check our other sons' newborn screening tests just to be sure. A few months passed before we were able to head in for another face-to-face in the hospital thanks to Covid, but when we finally got in to see the doctors, they assured us that they had been in contact with the relevant department in Sydney and our other two sons' tests were perfectly fine.

As Nicholas started solids, we were placed on a strict low-protein diet of just 6 grams per day which was slowly calibrated to include more protein as he grew.

By one year old he was allowed 12 grams of protein but no meat products or dairy. This was a strange situation for us as we knew Cobalamin C was treated a lot differently in the US, but we decided that we just had to trust in our doctors as Nicholas seemed to be doing really well.

In January 2021 we finally received Nicholas' genetic results confirming he did in-fact have cobalamin C disorder. This again raised concerns for me surrounding my other sons, and we asked if we could have them tested for CblC. Our doctors weren't very keen and explained testing them could have moral repercussions as it should be their decision. I kept pushing, and by October 2022, the hospital finally agreed to do a urine test to check for MMA on our other two sons. Again, we were reassured that they had double-checked their old screening tests which were all fine. We finally did the urine test on a Friday evening. Our sons did seem perfectly fine at this stage, and I just wanted to have that confirmation; it was never that we suspected anything. Both boys were happy, healthy and thriving.

The following Monday I had dropped the boys at school and daycare and had just arrived at the shops ready to buy Connor, our middle son, some presents for his upcoming 4th birthday when I got a call from my husband. The hospital had called Daniel and told him we needed to bring Connor up to the hospital right away. I broke down in tears in the middle of the shopping centre and I think this scenario was even more shocking for us than when we first found out about Nicholas. We had no reason to suspect that anything was wrong.

On 28th October 2022, just 3 days before his 4th birthday, it was confirmed that Connor also had Cobalamin C deficiency. We were told that our two sons represented 2/3 of Queensland's population of Methylmalonic Acidemia and there was just one other case in the whole population of Queensland.

Connor started his first year of formal education at school this year and it's going really well. We had concerns about the potential for any damage that may have been done while he was not being treated, but so far, his vision is fine, and he has no delays. Our son Nicholas is also doing really well and meeting his goals.

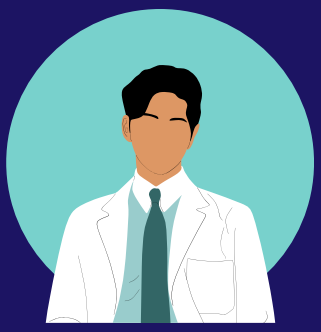
By sharing our story, I would really like to highlight how important it is for screening siblings of familial cases. It was a real effort for us to achieve this here in Australia and **I'm SO thankful that we never gave up on pushing their case.**

To read Nicholas and Connor's story on our website, visit
https://hcunetworkamerica.org/cobalamin-patient-stories/nicholas-connor-australia

Rare-X Data Collection Program

75 enrollees are on the platform, yet more are needed!

With permission, your deidentified data can help



doctors and researchers to find missing data to....

- Better understand homocystinuria
- Provide better treatment
- Develop new and better therapies
- FIND A CURE!

Enroll today at: https://homocystinuria.rare-x.org

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

UPDATE

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

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

What is the purpose of this program?

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

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

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

MEDICAL ASSISTANCE

Who is eligible to apply?

This program is designed to help patients who:

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

What is the application process?

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

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

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

NORD is Here for You

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

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

Alone we are rare. Together we are strong.®

How do I get more information and apply?

Contact NORD's Classical HCU Medical Assistance Program

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

203-616-4327

203-635-4163

hcu@rarediseases.org

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

What assistance does NORD provide?

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

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

Awards are issued for a calendar year.

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

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

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

How does NORD demonstrate compliance with regulations required of charities?

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

NORD®

rarediseases.org

Buffalo Jack'n

Will be demoed at our cooking class on 6/4!

Makes 3 servings | Serving size 86 g | 1.4 g protein or 61 mg PHE

Ingredients:

- 1 20-oz. can Jackfruit, canned, drained, seeds removed
- 1/2 c Mayonnaise
- 1/2 tsp Garlic Powder
- 1/4 tsp Onion Powder
- 1/4 tsp Salt
- 1/4 tsp Black Pepper
- 1 TBSP Buffalo Sauce or more to your liking

Directions:

1. Prep the jackfruit by rinsing well with cool water to remove the excess brine. Remove seeds, then squeeze out the excess liquid. Coarsely chop to resemble small chicken chunks. Add to a medium bowl.

2. Combine all other ingredients with the chopped jackfruit. Mix well to combine. Serve immediately or refrigerate until ready to use.

UPCOMING EVENTS

Find all events at: https://hcunetworkamerica.org/virtual-meet-ups/

Low Protein Cooking Class

June 4 | Time: 2 PM EDT

Will YOU be our next HCU HERO?

Tell us:

- How you or your child was diagnosed?
- How has HCU affected you, your family and relationships?
- What are some of your successes with HCU
- What are some of your challenges you have faced?
  - How have you overcome them?
- What words of advice would you give to newly diagnosed families?
- For other patient stories, visit: https://hcunetworkamerica.org/patient-stories
- Email your story to: info@hcunetworkamerica.org

World Homocystinurias Awareness Day - recap!

Raffle Winners

Caretaker's Care package: Samantha Mozdzen

The 3 Below basket: Kristin Rapp

The Chocolate, Cookies, and Crunches Basket: Ethan Hunt

Cold Brrrrew Crate: Jacqueline Piccini

Java Junkies: Samantha Mozdzen

$150 gift card: June Anderson

A special thank you to Dean Seppelfrick, Aimee Marchese w/Stressless Gifts, Starbucks & Synlogic for providing our awesome prizes!

WE WANT YOU!

JOIN OUR FUNDRAISING TEAM

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

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

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

Email us:
dbartke@hcunetworkamerica.org

SEPTEMBER 1-30, 2023

HCU HEROES

RACE FOR RESEARCH

WALK / RUN / RIDE

More swag available for fundraisers!

Per Individual: $30

Per Family (up to 4 – 1 mailing address): $75

Start your journey now: https://charity.pledgeit.org/HCURaceforResearch

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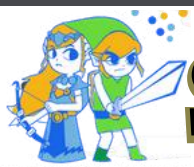
HCU NETWORK AMERICA

HCU HEROES

RACE FOR RESEARCH

WALK / RUN / RIDE

HEROES IN THE MAKING



SEPTEMBER 1-30, 2023

HCU NETWORK AMERICA

aeglea

What is a virtual race?

A virtual race is a race that can be walked, ran, or biked from any location you choose. You can participate on the road, on the trail, on the treadmill (or stationary bike), at the gym or on the track (or even at another race). You get to run your own race, at your own pace, and time it yourself. You do not have to complete the miles all at once, in one day, or even a week. You can use the entire month to complete the race.

How do you know how many miles I completed?

- Our preferred method to track your miles is Strava. When you sign sign up it will prompt you to connect.
- If you choose not to connect with Strava, you can manually enter your miles.
- Please log all your miles by 11:59 pm ET September, 30, 2023

How do my miles translate to money raised?

After a racer is registered, they are set up with their own personal donation page. You can direct those who would like to donate to your race link.

Learn more or register at https://charity.pledgeit.org/HCURaceforResearch

New Resources

Let's help you *Transition to Adulthood*!

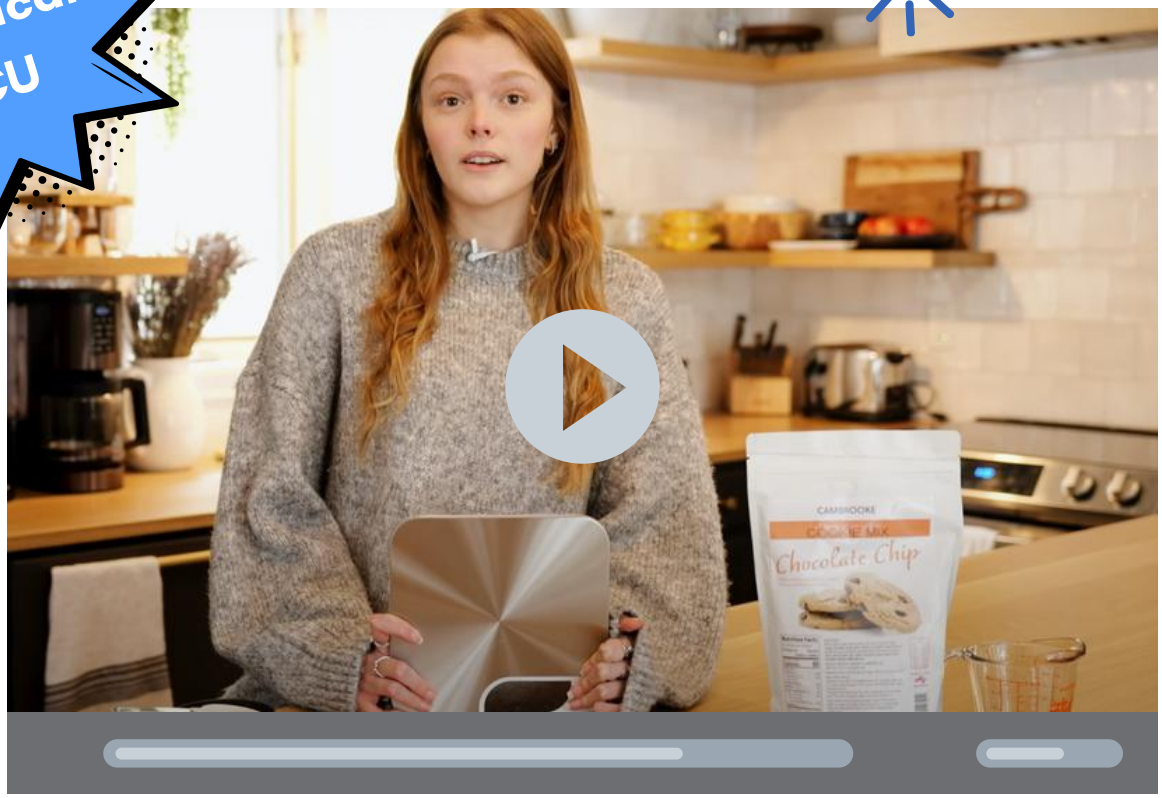
We're adding more resources to supplement our *Transition to Adulthood* milestones guide!

Medications and Medical Nutrition

I can accurately track my protein intake for the day.

Classical HCU

Click video to watch!



THANK YOU!

***to Gabbi**, **Chloe** and **Brendan** for creating this amazing resource for our community*!

Click **here** to access our *Transition to Adulthood* guide & additional resources!

New Resources

Let's help you *Transition to Adulthood*!

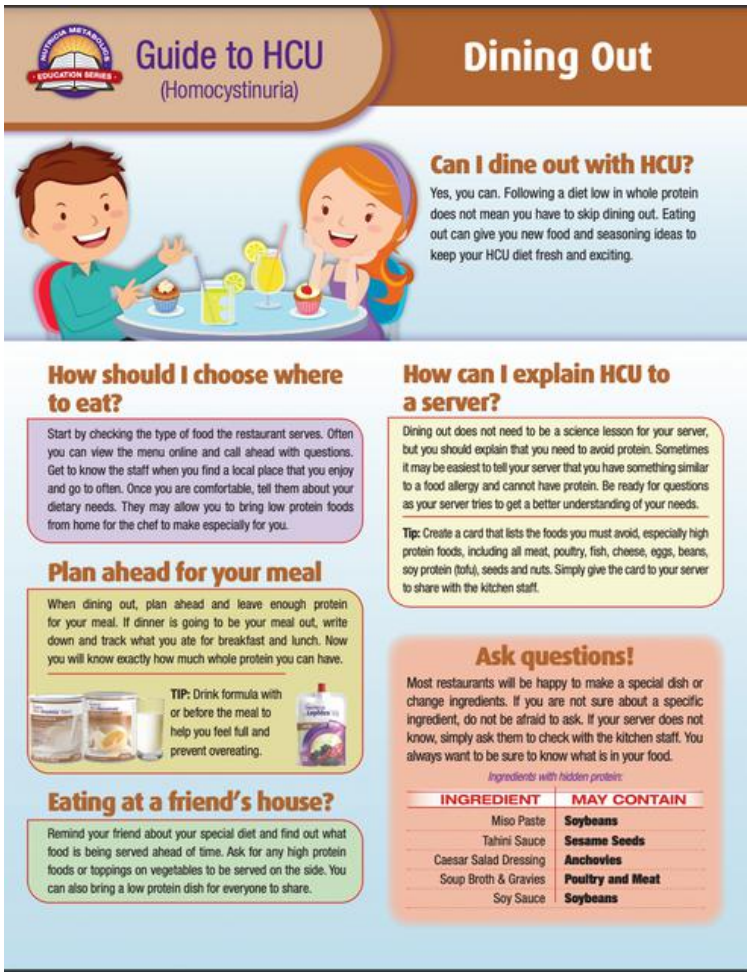
We're adding more resources to supplement our *Transition to Adulthood* milestones guide!

Medications and Medical Nutrition

I can make safe food choices when I'm away from home.

CLICK HERE

to download this resource



Guide to HCU (Homocystinuria)

Dining Out

Can I dine out with HCU?

Yes, you can. Following a diet low in whole protein does not mean you have to skip dining out. Eating out can give you new food and seasoning ideas to keep your HCU diet fresh and exciting.

How should I choose where to eat?

Start by checking the type of food the restaurant serves. Often you can view the menu online and call ahead with questions. Get to know the staff when you find a local place that you enjoy and go to often. Once you are comfortable, tell them about your dietary needs. They may allow you to bring low protein foods from home for the chef to make especially for you.

Plan ahead for your meal

When dining out, plan ahead and leave enough protein for your meal. If dinner is going to be your meal out, write down and track what you ate for breakfast and lunch. Now you will know exactly how much whole protein you can have.

TIP: Drink formula with or before the meal to help you feel full and prevent overeating.

Eating at a friend's house?

Remind your friend about your special diet and find out what food is being served ahead of time. Ask for any high protein foods or toppings on vegetables to be served on the side. You can also bring a low protein dish for everyone to share.

How can I explain HCU to a server?

Dining out does not need to be a science lesson for your server, but you should explain that you need to avoid protein. Sometimes it may be easiest to tell your server that you have something similar to a food allergy and cannot have protein. Be ready for questions as your server tries to get a better understanding of your needs.

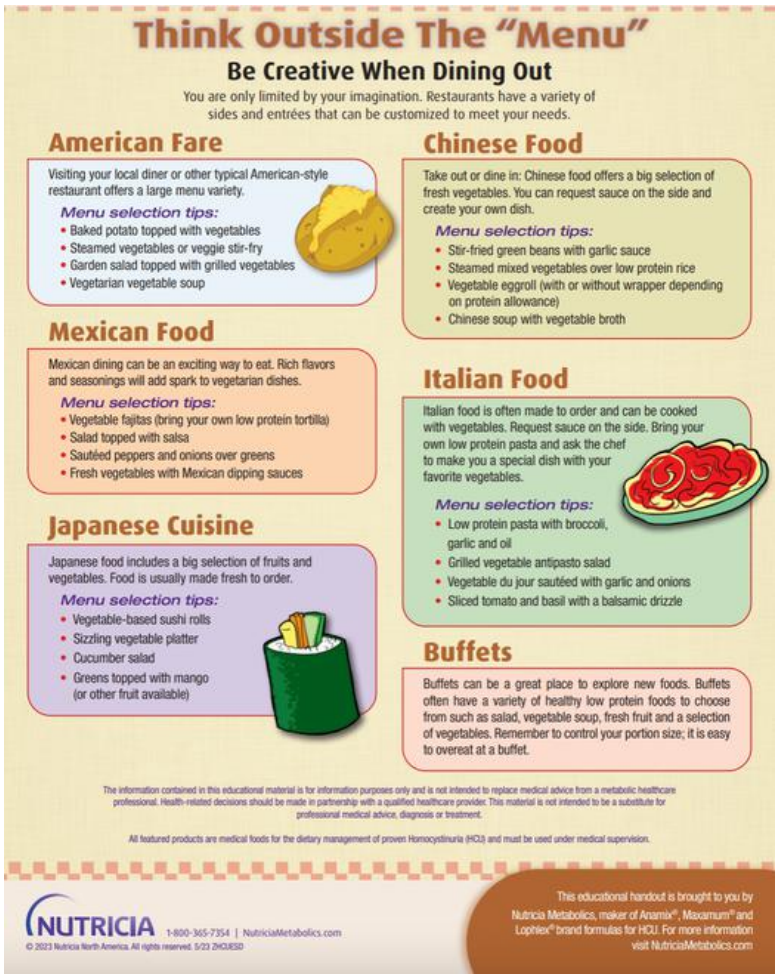
Tip: Create a card that lists the foods you must avoid, especially high protein foods, including all meat, poultry, fish, cheese, eggs, beans, soy protein (tofu), seeds and nuts. Simply give the card to your server to share with the kitchen staff.

Ask questions!

Most restaurants will be happy to make a special dish or change ingredients. If you are not sure about a specific ingredient, do not be afraid to ask. If your server does not know, simply ask them to check with the kitchen staff. You always want to be sure to know what is in your food.

Ingredients with hidden protein:

INGREDIENT	MAY CONTAIN
Miso Paste	**Soybeans**
Tahini Sauce	**Sesame Seeds**
Caesar Salad Dressing	**Anchovies**
Soup Broth & Gravies	**Poultry and Meat**
Soy Sauce	**Soybeans**



Think Outside The "Menu"

Be Creative When Dining Out

You are only limited by your imagination. Restaurants have a variety of sides and entrées that can be customized to meet your needs.

American Fare

Visiting your local diner or other typical American-style restaurant offers a large menu variety.

Menu selection tips:

- Baked potato topped with vegetables
- Steamed vegetables or veggie stir-fry
- Garden salad topped with grilled vegetables
- Vegetarian vegetable soup

Chinese Food

Take out or dine in: Chinese food offers a big selection of fresh vegetables. You can request sauce on the side and create your own dish.

Menu selection tips:

- Stir-fried green beans with garlic sauce
- Steamed mixed vegetables over low protein rice
- Vegetable eggroll (with or without wrapper depending on protein allowance)
- Chinese soup with vegetable broth

Mexican Food

Mexican dining can be an exciting way to eat. Rich flavors and seasonings will add spark to vegetarian dishes.

Menu selection tips:

- Vegetable fajitas (bring your own low protein tortilla)
- Salad topped with salsa
- Sautéed peppers and onions over greens
- Fresh vegetables with Mexican dipping sauces

Italian Food

Italian food is often made to order and can be cooked with vegetables. Request sauce on the side. Bring your own low protein pasta and ask the chef to make you a special dish with your favorite vegetables.

Menu selection tips:

- Low protein pasta with broccoli, garlic and oil
- Grilled vegetable antipasto salad
- Vegetable du jour sautéed with garlic and onions
- Sliced tomato and basil with a balsamic drizzle

Japanese Cuisine

Japanese food includes a big selection of fruits and vegetables. Food is usually made fresh to order.

Menu selection tips:

- Vegetable-based sushi rolls
- Sizzling vegetable platter
- Cucumber salad
- Greens topped with mango (or other fruit available)

Buffets

Buffets can be a great place to explore new foods. Buffets often have a variety of healthy low protein foods to choose from such as salad, vegetable soup, fresh fruit and a selection of vegetables. Remember to control your portion size; it is easy to overeat at a buffet.

THANK YOU!

*to **Nutricia** for providing this resource to our community!*

Click **here** to access our *Transition to Adulthood* guide & additional resources!

New Resources

Let's help you *Transition to Adulthood*!

We're adding more resources to supplement our *Transition to Adulthood* milestones guide!

Insurance

I know how to identify the important details on my insurance card (name of the policyholder, policy number, group number, etc).

CLICK HERE

to download this resource

I know how to identify the important details on my insurance card

As you start to become independent, it is important to carry a copy of your own health insurance card and to be able to identify the important parts. Your card may look different from the one shown but should have the same type of information. If you share a policy with your parents you will need to ask them to help you obtain a copy to keep for yourself. You will need this for every doctor's appointment, so please keep in your purse or wallet.

1. **Insurance company name:** This is the company that you have your insurance policy with. The company may be referred to as a carrier.
2. **Coverage or Plan type:** Your card might have a label like HMO, or PPO, to describe the type of plan you have. Knowing the type of plan will help you determine your network - you want to make sure your providers are in-network!
3. **Policyholder, subscriber name, member name:** This is the primary person, either yourself or a parent who has purchased or owns the insurance policy.
4. **Member number, member ID identification number, name ID, ID:** This number is specific to you - regardless of your plan within that company. It tells your healthcare provider how to bill your insurance plan.
5. **Group number:** This number is unique to your plan and is used to track the particular benefits your plan includes. If you get your insurance from an employer, all employees will have the same group number.
6. **Policy name:** Insurance companies offer a lot of different policies. This will be important to know, in understanding if your provider is covered and at what rate.
7. **Co-pay (copayment):** These are the amounts that you will owe when you get healthcare with the various types of providers.
8. **Pharmacy (Rx) benefits:** If you have pharmacy benefits with your healthcare plan, they will be listed like this. Some companies may provide you with a separate card.
   - **Rx BIN:** Tells the pharmacy the name of your insurance company.
   - **Rx PCN:** Helps the pharmacy locate your pharmacy member profile.
   - **Rx GRP, Rx Group:** Informs the pharmacy of your prescription benefits.

Tip!
Save a digital copy of your card to the notes section of your phone, or download your insurance company's app to your phone and create an account so you always have a copy of your card on hand!

HCU NETWORK AMERICA

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INDUSTRY NEWS

Travere Therapeutics Announces Positive Topline Results from Cohort 6 in the Phase 1/2 COMPOSE Study of Pegtibatinase in Classical Homocystinuria

SAN DIEGO, May 31, 2023 (GLOBE NEWSWIRE) -- Travere Therapeutics, Inc. (NASDAQ: TVTX) today announced positive results from cohort 6 in the placebo-controlled Phase 1/2 COMPOSE Study of pegtibatinase, a novel investigational enzyme replacement therapy being evaluated for the treatment of classical homocystinuria (HCU). In this cohort, five patients were randomized in a blinded fashion to receive 2.5 mg/kg of lyophilized pegtibatinase or placebo twice weekly (BIW), with four patients assigned to the treatment group. In this highest dose cohort to date, treatment with pegtibatinase resulted in rapid and sustained reductions in total homocysteine (tHcy), with a 67.1% mean relative reduction in tHcy from baseline, as well as maintenance of mean tHcy below the clinically meaningful threshold of 100 µM, over weeks 6 to 12. To date in the study, pegtibatinase has been generally well-tolerated.

To read the full press release, visit https://bit.ly/3U5MdY9

TRAVERE™ THERAPEUTICS

INDUSTRY NEWS

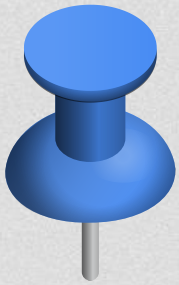
eTon PHARMACEUTICALS

Eton Pharmaceuticals Announces Commercial Availability of Betaine Anhydrous for Oral Solution, a Generic Version of Cystadane® (betaine anhydrous for oral solution)

DEER PARK, Ill., May 10, 2023 (GLOBE NEWSWIRE) -- Eton Pharmaceuticals, Inc (Nasdaq: ETON), an innovative pharmaceutical company focused on developing and commercializing treatments for rare diseases, today announced the commercial availability of Betaine Anhydrous for Oral Solution 180 grams for the treatment of homocystinuria.

“We are excited to be offering a lower cost alternative1 to Cystadane while still providing full patient and provider support services. We believe the adoption of our product will result in significant financial savings to the U.S. healthcare system and to many patients,” said Sean Brynjelsen, CEO of Eton Pharmaceuticals.

To read the full press release, visit [bit.ly/3pCZ365](bit.ly/3pCZ365)



Eton Cares patient support program offers a $0 co-pay to eligible, commercially insured patients!

Learn more here: https://www.betaineus.com/

Now Available!

Betaine Anhydrous for Oral Solution 180 gm

An AB rated Generic version of Cystadane®
(betaine anhydrous for oral solution) with full
patient support services you might expect from a Brand†

Copay support

Eton Cares can help eligible, commercially insured patients get their medication for **as little as $0 per month***

Financial support

Patients who do not have insurance and meet certain financial requirements may be eligible for additional financial support from our **Patient Assistance Program***

*Restrictions, limitations, and/or eligibility requirements may apply. For patients who are not elibile for copay support or who need additional financial assistance, Eton Cares can help connect you with alternative forms of medication coverage or provide referrals to other possible sources of funding.

Eton Cares can provide copay and financial support.

Have your doctor complete the referral form to prescribe and enroll.

Click Here

IMPORTANT SAFETY INFORMATION

Warnings and Precautions

Hypermethioninemia in Patients with CBS Deficiency: Betaine Anhydrous may worsen high methionine blood levels and accumulation of excess fluid in the brain has been reported. If you have been told you have CBS deficiency, your doctor will be monitoring your methionine blood levels to see if changes in your diet and dosage are necessary.

Adverse Reactions

Most common side effects were nausea and gastrointestinal distress, based on a survey of doctors.
To report a suspected adverse event related to Betaine Anhydrous, contact Eton Pharmaceuticals, Inc. at 1-855-224-0233 or the U.S. Food and Drug Administration (FDA) at **http://www.fda.gov/MedWatch** or call 1-800-FDA-1088.

INDICATIONS AND USAGE

Betaine anhydrous for oral solution is indicated in children and adults for the treatment of homocystinuria to decrease high homocysteine blood levels. Homocystinuria is a rare genetic disorder in which there is an abnormal accumulation of the amino acid homocysteine in the blood and urine. The following are considered to be homocystinuria disorders:

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

eTon PHARMACEUTICALS

Please see enclosed Full Prescribing Information for more information.

†Cystadane is a registered trademark of Recordati Orphan Drugs SAS, not affiliated with Eton Pharmaceuticals.

1402-v2

Acappella

NOW ENROLLING

ACAPPELLA Study on Classical Homocystinuria

Travere Therapeutics is enrolling children and adults with classical homocystinuria (HCU) in a ACAPPELLA Study. The goal is to learn more about classical HCU and the 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 participants.

Approximately 150 participants will take part 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 ACAPPELLA Study if you:

- **Have been diagnosed with HCU**
- **Are 1–65 years of age**

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

For additional information about the ACAPPELLA 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 ACAPPELLA Study.
Sites are open and currently enrolling participants.

For new participants, we now have an option for the study to come to you! (decentralized site).
Please inquire to learn more.*

If you have any questions, please email:

medinfo@travere.com

For more information, please scan the QR code or visit www.hcuconnection.com

*Restrictions apply

MA-PE-22-0004. March 2023

TRAVERE THERAPEUTICS

Contact Register

What is the contact register?

The contact register is a secured private survey that allows you to share information on you or your family member with HCU with us. This includes where you are from, your relationship to homocystinuria, the patient's birthdate, gender, their exact diagnosis (e.g. CBS, cobalamin, or MTHFR), how they were diagnosed, and if the patient was diagnosed through newborn screening. This information is kept confidential and will not be shared unless you give us permission. By registering, you will be able to identify other patients in your state and request their contact information. You will also be able to access information posted over time that can only be shared with the patient community. (For example, we may have webinars that the expert presenter does not want to be publicly available, but is willing to share with the HCU community.)

What will this information be used for?

HCU Network America strives to inform patients and families with resources, create connections, and support advancement of diagnosis and treatment of HCU and related disorders. The information you provide helps us succeed in our mission – plan events, develop resources and educational tools, and ensure everything is being done to support timely and accurate diagnosis from birth. It also allows us to have informed conversations with doctors, pharmaceutical companies, and law makers. Your information helps us understand the landscape better so we can better advocate for you!

How do I participate?

The contact register form takes approximately 3-5 minutes to complete. You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to: https://www.surveymonkey.com/r/HCUContact

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