CLASSICAL HOMOCYSTINURIA

A toolkit for managing cystathionine beta synthase

HCU NETWORK AMERICA

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WELCOME

Whether diagnosed at birth, as a child or as an adult, Homocystinuria comes as a shock to everyone. Adjusting to a diagnosis comes with uncertainty, confusion and a huge learning curve. Know that all the feelings you are experiencing are normal and natural. Other patients and parents before you have felt these same emotions you are experiencing.

When my brother and I received our diagnosis when I was 10, we were very uncertain about our future. The data at the time had painted a very bleak picture. You see, in 1995, there weren't any FDA approved treatments, clinical trials, and absolutely no research happening.

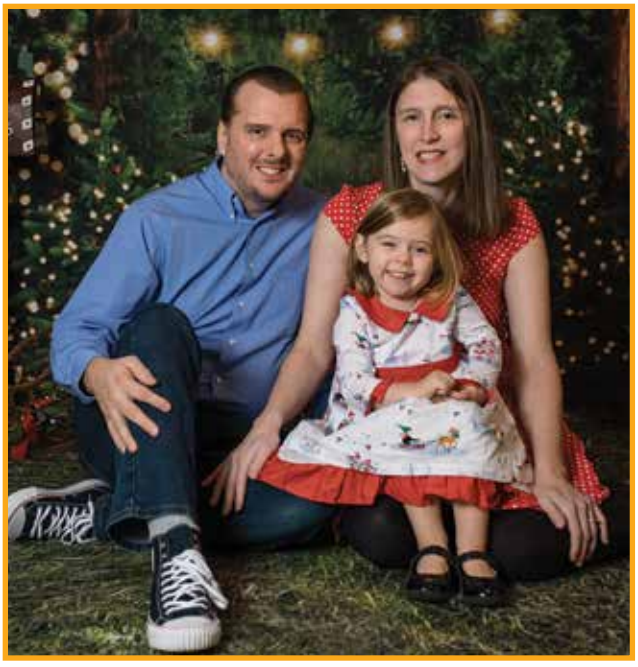
The Natural History Study done in the 1980s showed patients on average lived to the age of 30, and female patients should never entertain the idea of having their own family. To add to the uncertainty and growing concern we felt very isolated and alone with our diagnosis. The internet hadn't quite evolved to the point it has today making it virtually impossible to connect with other patients and families with Homocystinuria.

But there is good news – things have advanced, and you are not alone in your days and years ahead.

I am now an adult past that 30-year-old milestone and have a family of my own. Patients are getting diagnosed earlier through newborn screening and awareness initiatives through clinics. We now have newborn screening in every state as well as growing treatment options available. With new research and clinical trials for potential new therapies, we now have an even better understanding of the disease. While these things happen and evolve, consider HCU Network America a friend and resource that can help provide comfort and guidance along the way. With that support and the tools we provide, things will get easier.

Our mission at HCU Network America is to help patients and caregivers affected by HCU and related disorders manage their disease and ultimately find a cure. If you look over our toolkit, you will get an overview of the information we provide showing the stages of development and expectations from infancy to adulthood. Our website, https://hcunetworkamerica.org, includes additional information on managing Classical HCU, ongoing research, patient stories, and much more.

Depending on where you are in your journey, things may seem overwhelming as you learn to navigate living with HCU; however, with community support and our resources you will be on your way to success!



Know that with time, patience, hard work as well as having good communication with your physicians and your new friends at HCU Network America – everything will be okay!

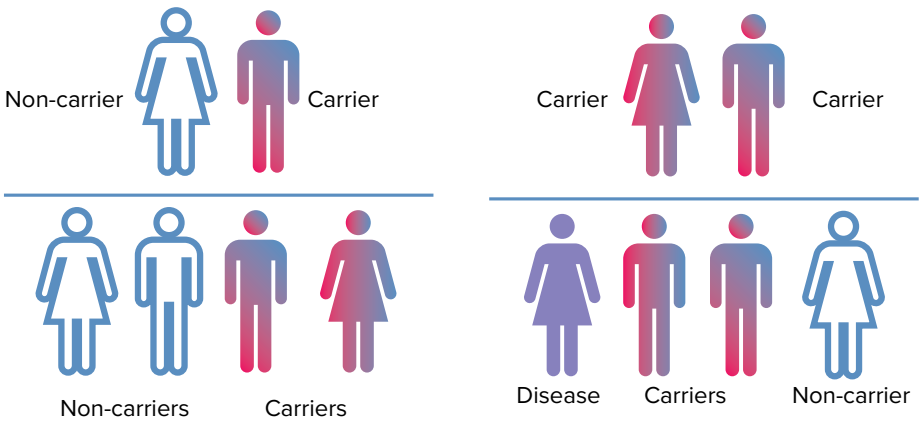
Sincerely Yours,
Danae' Bartke
Patient and Executive Director
HCU Network America

PART 1: WHAT IS HOMOCYSTINURIA?

Homocystinuria (HCU) is a congenital disorder affecting the metabolism of amino acids. Classical HCU is caused by the mutation of a gene that codes for Cystathionine Beta Synthase (CBS), the enzyme that metabolizes homocysteine into cystathionine, another amino acid. The absence of CBS prevents this reaction and causes elevated homocysteine, which can cause ophthalmic, circulatory, CNS and skeletal issues. If identified at birth, early treatment can enable normal development and helps decrease the chances of damage.

Classical HCU is an autosomal recessive disease, meaning each parent needs to be a carrier and contribute to the defective gene in the child.

Statistically, 1 out of 4 children of carrier parents would be expected to have the disease, with 2 out of 4 becoming carriers.



THE ROLE OF METHIONINE IN HCU

Amino acids like methionine are the building blocks of proteins; methionine is an essential amino acid. This means that the body cannot make it on its own methionine and it must be provided through food. Methionine is critical in making common proteins. Homocysteine is also converted back into methionine. Consuming methionine can be problematic for those with Homocystinuria.

HOW COMMON IS HCU?

HCU is estimated to affect 1 in 200,000-355,000 people in the U.S., but higher in some countries. Ireland, Germany, Norway and Qatar have some of the highest incidences of Homocystinuria.

Ireland	1 in 65,000
Germany	1 in 17,800
Norway	1 in 6,400
Qatar	1 in 1,800

WHAT AREAS OF THE BODY ARE AFFECTED?

Eyesight is one of the most affected abilities. Many people with HCU are extremely nearsighted. Unfortunately, their vision often gets worse with time if not diagnosed or noncompliant with their treatment plan. Bone density and shape is also a concern. Regular bone screenings are recommended.

CIRCULATORY SYSTEM
Strokes
Blood Clots
(B6 responsive patients may be asymptomatic or only suffer adult onset thromboembolism)

CENTRAL NERVOUS SYSTEM
Psychiatric & Behavior Disorders
Learning Difficulties
Intellectual Disability
Movement Disorders
Seizures

SKELETAL SYSTEM
Excessive Height
Long Limbs
Osteoporosis
Bone Deformities
Scoliosis
Knock knees
Pigeon Chest

EYES
Lens Dislocation
Nearsightedness
(Myopia)

MONITORING AND RECOMMENDATIONS

The following tests and assessments are recommended. Please consult your doctor or specialist for more information.

AREA	TESTS	FREQUENCY
Anthropometry	Height & Weight	Every clinic visit
Dietary	Dietary intake analysis	Every clinic visit of on dietary treatment
Biochemical-metabolic control	tHCY, Met	Semi-Annually or annually, unless determined otherwise more frequently by medical team
Nutritional	Vitamin B12, folate, blood count, albumin, plasma AA, ferritin, zinc, 25-hydrovitamin D	At least annually if on dietary treatment
Neurodevelopmental/neurological	Clinical examination | MRII/EEG	Annually | Only if new CNS symptoms
Opthalmologic	Eye examination	At least annually
Neuropsychological fluctuations	IQ test	At least every 5 years in childhood
Psychological	Clinical psychology or psychiatric assessment	As required
Bone density	DEXA	Every 3-5 years from adolescence unless clinically indicated earlier
Cardiovascular	Lipid profile, cardiovascular risk factor review	Once in childhood; annually in adulthood

HOW IS HCU DIAGNOSED?

All 50 states screen for HCU at birth. Homocystinuria (CBS Deficiency) can be confirmed by mutation analysis or CBS assay. Because HCU shares many similar skeletal and eye features with Marfan's Syndrome, people with HCU are mistakenly diagnosed as having Marfan's Syndrome.

NEWBORN SCREENING

Since 2007 all 50 states have screened for HCU at birth via newborn screening. Unfortunately though, at least 50% of cases are not picked up by newborn screening. These 50% of patients are often but not always those who have the milder, vitamin B6 (pyridoxine) responsive or partially vitamin B6 responsive form.

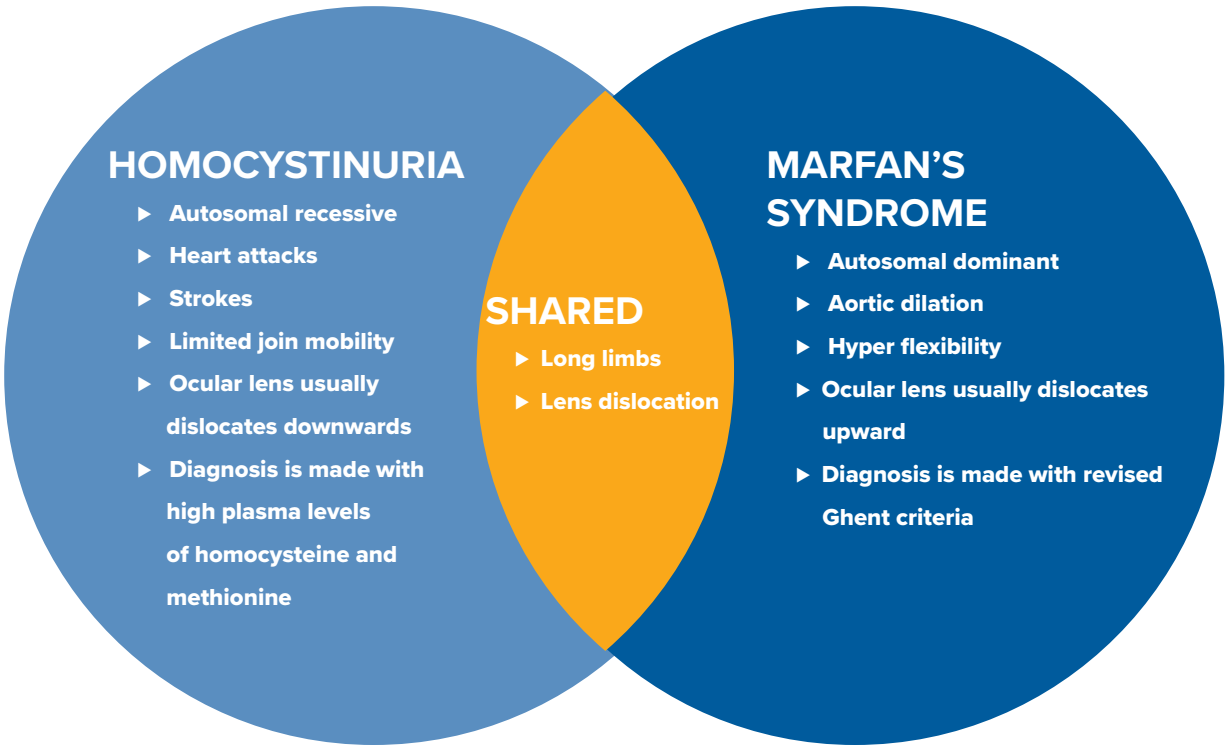
- The newborn screening test uses a heel prick to measure methionine levels – if high, homocysteine levels are then measured.

LATE DIAGNOSES

- If missed by newborn screening, HCU can be diagnosed later due to clinical symptoms. It takes an average of 4.5 years from first symptoms to receive an accurate diagnosis.
- By 10 years of age, without treatment 55% of B6-responsive and 82% of non-responsive will have lens dislocation.
- By 15 years without treatment 12% of B6-responsive and 27% of non-responsive will have a thromboembolic event.

When testing for HCU doctors should use the plasma total homocysteine test since testing free homocystine is unreliable.

- Patients should not be taking a multivitamin or vitamin B6 prior to the blood test.
- Plasma should be separated promptly.



WHAT IS THE CLINICAL PRESENTATION?

In a healthy person who is not affected by HCU, plasma homocysteine usually will not exceed 15 (umol/L) of blood. Healthy carriers may have less ability to metabolize homocysteine and have elevated levels that exceed 15umol/L.

If you are a carrier with heightened levels, please seek the advice of a metabolic specialist.

Patients on treatment may have levels greater than 15 (umol/L), untreated patients often have levels several fold higher.

B6 (PYRIDOXINE) RESPONSIVE OR NOT

HCU patients can be broken down into two categories, B6 responsive or non-responsive.

B6 RESPONSIVE

About 50% of patients are vitamin B6 (pyridoxine) responsive or partially vitamin B6 responsive. A few patients who are vitamin B6 responsive do not require a low-protein diet, but most still require some restriction of protein and/or the HCU formula. If patients cannot reduce their homocysteine levels below 50 umol/L with B6 alone, they may require the addition of betaine to their daily regimen.

B6 NON-RESPONSIVE

The other half of patients are considered B6 non responsive. B6 non-responsive patients are usually on a low-protein diet, often with betaine added, and require specialty low-protein medical foods and supplements of other amino acids. The goal for these patients is to reach levels below 100 umol/L, though some physicians will recommend lower levels if they can be reached safely.

PROGNOSIS

Early diagnosis and adherence can prevent the onset of the symptoms. Adherence to the medication and diet must be constantly monitored. If the homocysteine level is kept below 70-100umol/L, cognitive and developmental delays, as well as the chance for blood clots and strokes can be greatly minimized. Some clinics may aim for lower levels.

MONITORING

Frequency of clinic visits and lab tests are dependent upon severity, treatment and age.

Total homocysteine and methionine values should be monitored and discussed at each visit.

In addition, patients should periodically have a folate, vitamin B12, full amino acid panel and bone density scan.

WHAT CAUSES HIGH LEVELS?

Multiple factors can play a role in patients' homocysteine and methionine concentrations.

- Endogenous Inborn Errors of the Metabolism
- Excessive protein intake, resulting in increased methionine
- Lack of CBS enzyme activity
- Vitamin B12 or folate deficiency
- Methylation disorder (e.g MTHFR)
- Sickness or stress (any kind), resulting in catabolism – the breakdown of muscle that releases amino acids into the blood for energy

PART 2: HCU NUTRITIONAL GUIDE

Because there is no cure for HCU, people with homocystinuria need life-long treatment. One of the most effective ways to manage HCU is through diet. Because patients with HCU have elevated levels of methionine and can't break down homocysteine, they need to limit the amount of methionine they ingests. Methionine is an amino acid found in foods like chicken, fish, eggs, and milk.

LOW METHIONINE/ PROTEIN DIET

Methods for calculating intake vary among metabolic clinics. Some recommend that patients who require a low-protein diet count methionine intake from food (in milligrams), while other clinics recommend that patients count overall protein intake (in grams).

The intake goal will vary depending on the patient's homocysteine and methionine levels. Both methods require careful measurement of food. Patients will get most of their protein from fruits, vegetables and synthetic low-protein foods, which include low-protein pasta, bread, rice, dairy, egg, dessert and meat substitutes.

HCU MEDICAL FORMULA

An important component for success to those on a low-protein diet is HCU medical formula. Without HCU formula, it can nearly be impossible to meet total protein requirements for normal growth and development. Formula provides all amino acids (except methionine) and the majority of vitamins and minerals a person needs while on a low-protein diet.

**Please see our Medical Formula and Low-protein vendor list for additional resources*

MEDICATION

Vitamin B6 is given to B6 responsive patients to stabilize any remaining CBS enzyme. If folate (B9) or vitamin B12 (Cobalamin) levels are low, elevated homocysteine may result, so many patients take low doses of B12 and folate.

For patients who cannot reduce homocysteine to the targeted range, they may require the addition of Betaine, a supplement that helps convert homocysteine back to methionine. Betaine dosage depends on the level of (if any) active enzyme, height, weight and age.

Your health care provider may have you add other vitamins or medication to ensure your health is optimized.

THE IMPORTANCE OF DIET

A low protein diet with medical food is paramount for management of classical homocystinuria (HCU). Good metabolic control cannot be achieved without restricting methionine, and methionine-free formula is essential for meeting total protein and micronutrient requirements for growth in childhood/adolescence, and maintenance health in adulthood. The diet is hard and often frustrating to follow, but major advances in the medical food industry over recent years have significantly improved the taste and nutritional quality of low protein foods and formulas. All patients report feeling better while on diet, and significantly reduce their risk for HCU-related medical complications.

– Angela Dempsey, RD, HCU Network America Medical Advisor

BASIC RULES OF THE DIET

The dietary regimen has some ground rules, the most important being to keep plasma Homocysteine levels within safe limits.

LIMITING METHIONINE

Methionine is found in all natural proteins. Because methionine is an essential amino acid, everyone needs some methionine in their diet. For patients with HCU, providing just enough methionine to meet needs for growth and/or maintenance is the key. Calories from other foods, including low-protein food/ medical formula, are very important and come from the following sources:

- Foods naturally lower in protein, like fruits and vegetables, are recommended as the staple of the diet regime.
- Special low-protein foods such as flour, bread and pasta.
- Measured amounts of regular grains, such as rice and crackers
- Special low-protein food and medical formula
- All patients should be followed by a metabolic dietitian who can help personalize their diet therapy.

BALANCING THE DIET

Note that the dietary guidelines for HCU do NOT include high protein foods such as meat, fish, eggs, soy, nuts and even some high-protein grains. Patients must make sure they are consuming adequate amounts of vitamins, minerals, carbohydrates and essential fatty acids for their age, sex and energy needs.

Consulting a dietitian will help you find the right medical formula and natural protein combination. With their advice, patients will be able to develop and grow at a normal rate while restricting their methionine intake.

RECOMMENDED PROTEIN LEVELS

A low-protein diet must always meet individual protein needs. Recommendations for total protein (protein from food, plus medical formula) vary with age and can be between 1 and 3 grams of protein per kilogram of body weight per day.

Work with your dietitian to find the right balance of total protein for you.

NUTRITIONAL NEEDS BASED ON AGE

As patients diagnosed with HCU age, their nutritional needs will change.

FEEDING BABIES WITH HCU

When HCU is diagnosed as a result of Newborn Screening, it is important to begin dietary treatment immediately.

Breast milk and standard infant formulas contain small quantities of methionine. Breast feeding babies with HCU is encouraged and helps create a strong tie between mother and child. Breastfeeding and infant formula may need to be supplemented with an appropriate HCU medical formula.

FEEDING CHILDREN WITH HCU

Children are growing and have high calorie and protein requirements.

A child's methionine/natural protein tolerance will dictate the dietary approach. A diet deficient in total protein would result in poor growth. There must be enough total protein on board to keep the child healthy and ensure growth. This cannot be accomplished without medical food.

Additionally, since methionine is an essential amino acid, it is rate-limiting, which means that too little natural protein from food will also result in poor weight gain and growth. Methionine intake cannot be "too much" or "too little" – it must be "just right."

While watching the protein or methionine content, it's also important to make sure the caloric needs are being met. Insufficient calories could compromise growth and lead to catabolism of protein from muscle, which would release methionine.

The diet should be monitored by a daily diet log and by blood draws. Your dietitian and geneticist will make adjustments from your diet log and your lab results

DIETARY NEEDS FOR TEENS AND ADULTS

The correct diet allows normal development of adolescents, mentally and physically, helping them lead normal lives.

The teenage and adult medical formula supplements are available in easy packaging to accommodate an active lifestyle.

Amino acid requirements for adults are not greatly different than that of children, but there are other nutrients that are just as important:

- Calcium and Vitamin D: to prevent osteoporosis and promote bone health
- Folic Acid: to prevent arteriosclerosis
- Fiber: for normal digestive health
- Omega 3 essential fatty acids: which strengthen immunity and have neuroprotective effects.

Metabolic clinic visits are less frequent in adulthood than in childhood, but regular monitoring is still important.

ANNISTON'S STORY

Anniston has been allowed 50 mg of methionine a day. She was recently increased to 75 mg. Baby food has been super easy since you know how much methionine is in each tub. Now that she's starting to eat more solids we are learning how to measure all her foods with a scale. We are learning more and more about what foods she can have and how to prepare them.

– Brittany, Mom to Anniston

PART 3: HCU AND PREGNANCY

It's especially important for women with HCU to take the proper precautions before and during pregnancy to ensure they have a safe and successful pregnancy.

CONSULT WITH YOUR TEAM

If possible, women should consult with their metabolic team before getting pregnant, about their decision to get pregnant. While pregnancy does pose some risk for women with HCU, it can be done in a very safe manner. It is likely you will be advised to consult with a hematologist and maternal fetal medicine specialist.

It is important that you have your metabolic team follow you closely when considering getting pregnant and during the pregnancy itself.

CLOSELY FOLLOW YOUR DIET

In the case of women who are not B6 responsive, it is also important that you closely follow your prescribed food and formula diet that your metabolic team has prescribed for you. As you progress in your pregnancy, your metabolic team will likely adjust your formula and protein intake to make sure you are meeting you and your growing baby's needs!

BIRTH CONTROL AND HCU

As previously stated, HCU comes with an increased risk for blood clots. Estrogen-based birth control, even in the general population, increases the chances for a blood clot. Because of this, it is highly recommended that women with HCU avoid estrogen-based birth control.

MEGAN'S STORY

Hi, my name is Megan Jackson and I am a mother of two sweet kiddos and I have HCU. I was beyond blessed by an amazing medical team, who all worked together so well to make sure each of my pregnancies went smoothly and I was safe.

While pregnant, any woman could have a blood clot or miscarriage, but as a woman with HCU my chances were even greater. I was closely monitored and in constant communication with my doctors, especially my genetic specialist.

Throughout my pregnancy, I got regular blood draws and kept a food diary, which I shared with my genetic specialist and dietician. They then would calculate where I was in my pregnancy, taking into account what my labs were and come up with a plan on how much protein, calories and protein supplement I should take. In the end, having my two kiddos is a true blessing.

PART 4: TIPS AND TRICKS

Don't get hung up on the details - here are a few tips and tricks to help you be more successful when grocery shopping, cooking, eating out and traveling.

SHOPPING AND COOKING

- Look at the food labels
- Double check the serving size and protein content for the listed serving.
- Generally, foods are weighed after they have been peeled or trimmed and before cooking.
- Ingredients in many high protein recipes can be swapped out for low-protein items.

EGGS

- For one egg, use ¼ cup unsweetened applesauce
- For one egg, use ¼ cup smashed banana
- For one egg, use ¼ cup low-protein plain yogurt
- Use Ener-g Egg Replacer – follow directions on box

MILK

- Cooking: Rice milk – ¾ cup for every 1 cup regular milk.
- Baking: Almond milk 1 cup for every cup.

FLOUR

- Low-protein baking mix – follow directions on box

CHEESE

- High protein cheese can be substituted for low-protein or some vegan cheeses.

EATING OUT

Having HCU doesn't mean the end of eating out. People and restaurants are more aware of food allergies and special diets than they have ever been! Keep these ideas in mind when you visit a restaurant or fast food joint.

- Keep a small food scale with you to weigh your items.
- Try to view their menu online in advance or call ahead to the restaurant.
- If you don't see something that suites your diet perfectly, don't be afraid to ask if they can substitute something.
- For example, substitute a Portobello mushroom for a beef hamburger patty.
- Some restaurants will let you bring your own low-protein pasta and cook it for you. Call ahead to see if they will cook or heat up your pasta for you.

TRAVELING

Exploration shouldn't be limited by HCU. Prepare yourself and give yourself plenty of time. This will help ensure that your travels will go smoothly and will be enjoyable. Here are some tips to help you on your way.

- Gather a note of medical necessity from your metabolic doctor
- This note should explain Homocystinuria, what medications, supplements, formula and food items you may be traveling with
- Check out the food scene where you are going; it may be necessary to pack your low- protein foods. Check out the Go Lo wPro App to assist you with this task!
- Pack extra food and formula, you never know when your flight might get delayed
- Carry as much of your medication, supplements and food as a carry on. Some airlines will allow an additional carry on if medically necessary. Check with your airline to see if they have such a policy
- Arrive at the airport with plenty of time. Because of the nature of supplements and formula, your luggage will likely get pulled aside and searched. Be patient and show them your letter of medical necessity. They will still check the contents, but it will make things move much smoothly!
- Have a written letter of what should happen in case of a medical emergency. Work with your medical team to complete this.
- Once on the airplane or in the vehicle, make sure you take plenty of stretch breaks to promote circulation.

ADAM'S STORY

With his desire to be normal, to overcome, to not be hampered by his medical condition, Adam wanted to try everything. He got hurt along the way, but that did not slow him down. No matter how many bumps or bruises Adam endured, he quickly bounced back, ready for the next new experience.... God made it very clear to me Adam was not to be boxed up or bubbled up. He is here for a purpose, and we need to let him go.

– Annettle Settle, Mom to Adam Settle

PART 5: FOOD AND FORMULA RESOURCES

MEDICAL FORMULA VENDORS

- **Abbot Nutrition**
  (800) 551-5838
  https://abbottnutrition.com/infant-and-new- moth-er#metabolic

- **Mead-Johnson**
  (812) 429-6399
  mjmedicalaffairs@mjn.com
  www.hcp.meadjohnson.com/products/ metabol-ic-products/

- **Nexus Patient Services**
  (833) 875-0200
  info@nexuspatientservices.com
  https://nexusmedicalnutrition.com/home

- **Vitaflo USA**
  (888) 848-2356
  VitafloNAM@VitafloUSA.com
  www.vitaflousa.com/

LOW-PROTEIN FOODS AND MEDICAL FORMULA VENDORS

- **Ajinomoto Cambrooke**
  (866) 456-9776
  info@cambrooke.com
  www.cambrooke.com/

- **Nutricia Metabolics**
  (800) 365-7354
  info@medicalfood.com
  www.nutriciametabolics.com

LOW-PROTEIN FOOD VENDORS

- **Dietary Specialty**
  (888) 640-2800
  http://dietspec.com

- **Lil's Dietary Shop**
  (773) 239-0355
  Lilsdietary.com

- **PKU Perspectives**
  (866) 758-3663
  Email: Sales@pkuperspectives.com Website: http:// pkuperspectives.com

- **Taste Connections**
  (310) 413-6499
  tasteconnect@verizon.net
  http://Tasteconnections.com

- **Zoia Pharma**
  (877) 379-9760
  Email: sales@zoiapharma.com
  https://www.zoiapharma.com/marketplace

OTHER NOTABLE FOOD COMPANIES

While these companies do not design things with our community in mind, many of their products will work for those on a low-protein diet. Please check the nutrition labels and consult with your dietitian if you are not sure if their products are right for you. Most of these companies can be found in most grocery stores or health food stores.

- **Better Than Foods**
  Pasta and rice alternatives
  (855) 691-5900
  Jeff@GreenSpotFoods.com
  www.betterthanfoodsusa.com/WebV31/

- **Daiya Foods**
  Dairy alternatives
  (877) 324-9211
  cr@daiyafoods.com website: https://daiyafoods.com

- **Ener-g Foods Company**
  Baking mixes, breads, and snack items
  (800) 331-5222
  customerservice@ener-g.com
  www.ener-g.com

- **Field Roast: Dairy Alternatives**
  (800) 311-9497
  http://fieldroast.com/product/chao-slices/

- **Follow Your Heart**
  Dairy-free alternatives
  (818) 348-3240
  amason@followyourheart.com
  http://followyourheart.com

- **Glutino**
  Baking mixes, snacks, breads, and breakfast pastries
  (800) 845-7286
  Website: http://glutino.com

- **The Jackfruit Company**
  Meat alternatives
  (877) 433-4024
  knowjack@thejackfruitcompany.com
  http://thejackfruitcompany.com

- **So Delicious**
  Dairy-free alternatives
  (866) 388-7853
  http://sodeliciousdairyfree.com/

- **Upton's Naturals**
  Meat alternatives
  (312) 666-7838
  info@uptonsnaturals.com
  uptonsnaturals.com

- **Violife Foods**
  Email: available through their website
  https://violifefoods.com/us

OTHER IMPORTANT COMPANIES

- **Recordati Rare Diseases**
  (Makers of Cystadane); Cystadane can be purchased by prescription through AnovoRx
  Recordati Rare Diseases' mission is to reduce the impact of extremely rare and devastating diseases by providing urgently needed therapies. We work side-by-side with rare disease communities to increase awareness, improve diagnosis and expand availability of treatments for people with rare diseases
  www.cystadane.com | (844) 288-5007

- **Anovo Rx**
  (888) 487-4703 | *www.anovorx.com*

- **Solace Nutrition**
  Solace Nutrition prides itself on its main reason for existence – the advancement of disease management through nutrition. Their products are designed by health care professionals specialized in the area of disease for which the products are intended.

  The Solace Nutrition team is consistently developing scientifically proven products that fill the unmet need of the many people struggling with inborn errors of metabolism and chronic disease management. Their products are in concentrated & convenient forms using only safe, clinically-validated ingredients that are manufactured in FDA registered & audited facilities.

  Products for HCU: VB6 P5PTM
  (888) 876-5223 | *www.solacenutrition.com*

LOW-PROTEIN FOOD AND MEDICAL FORMULA INSURANCE AND ASSISTANCE PROGRAMS

We realize that food and formula coverage varies greatly by state. For this reason, HCU Network America has contracted Raenette Franco of Compassion Works Medical LLC to help assist you in getting low-protein foods, formula and supplements covered. She will also work with you to find a policy that meets your individual needs if you don't currently have one or are struggling to find one.

Part of our mission as HCU Network America is to increase access for treatments and supplements for Homocystinuria patients. In order to fulfill this part of our mission, we have been in contact with Raenette Franco of Compassion Works Medical, LLC to help us fulfill this part of our goals.

MISSION OF COMPASSION WORKS MEDICAL

To change the lives of the rare genetic disease community by supporting a genuine helping hand with insurance coverage challenges. "We don't take NO for an answer along with compassion that makes all of us successful," says founder Raenette Franco.

Over the past years at Compassion Works Medical, Raenette has expanded her support to several special diseases that require medical foods and enteral nutrition, including cancer, ALS and TBI. "It is truly rewarding and my inspiration grows stronger and stronger to help those people find a way to afford and stay on diet," Franco says.

ABOUT FOUNDER RAENETTE FRANCO

A native New Yorker, Franco came to New Jersey in 2001 and worked as medical biller insurance specialist/consultant. Her background consists of hospice and palliative care, bariatric surgery, and other medical specialties related to insurance coverage. In 2011, Raenette landed a job within the medical food arena as a medical food insurance specialist. Inspired by helping others, Raenette founded Compassion Works Medical.

Raenette received her Certification as a Certified Biller Coder Specialist from the NCCA Accreditation in New Jersey. Her specialty is working directly with medical food coverage for both formula and low-protein foods. Raenette is dedicated to helping patients, dietitians, and physicians avoid the difficult tasks of insurance coverage and reimbursement issues for medical foods/enteral nutrition.

CONTACT RAENETTE FRANCO

Email: raenettef@compassionworksmrs.com
Phone: (973) 832-4736

OTHER ASSISTANCE PROGRAMS

Healthwell Foundation: Each year, more and more Americans are being forced to choose between paying for life-saving treatments and paying for basic necessities like food, housing and utilities – decisions no one should have to make. When people in these circumstances need help, many of them turn to the HealthWell Foundation.

Founded in 2003, the HealthWell Foundation is an independent, non-profit organization that provides financial assistance to insured patients living with chronic and life-altering illnesses to help them afford their medical treatments.

CONTACT HEALTHWELL FOUNDATION

(800) 675-8416
www.healthwellfoundation.org/fund/homocystinuria

DIET TRACKING APPLICATIONS AND WEBSITES

HCU Network America does not endorse any diet tracking application or website.

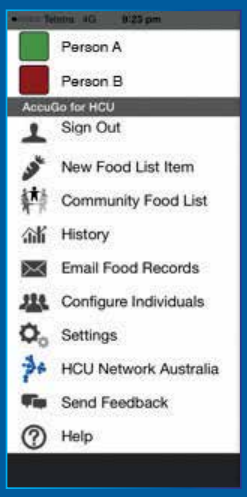
FOR MORE INFORMATION, CONTACT
HCU AUSTRALIA
Tara Morrison
President
+61 411 488 476
tara@hcunetworkaustralia.org.au
HCUNetworkAustralia.org.au

ACCUGO FOR HCU

AccuGo for HCU was designed to ease the day-to-day management of the low-protein diet by providing a quick and easy way to estimate and track protein or methionine in food eaten. It provides easy access to the protein and methionine content of foods including preloaded data, your own personal data and a community database of shared food.

"For my son's whole life, I have had to carry around my photocopied lists of food, a journal to keep track of what he ate, and a calculator to workout the protein value.

We now have a device solely used only for the app so that we can hand it off to any caregiver if needed. It's been a life saver."



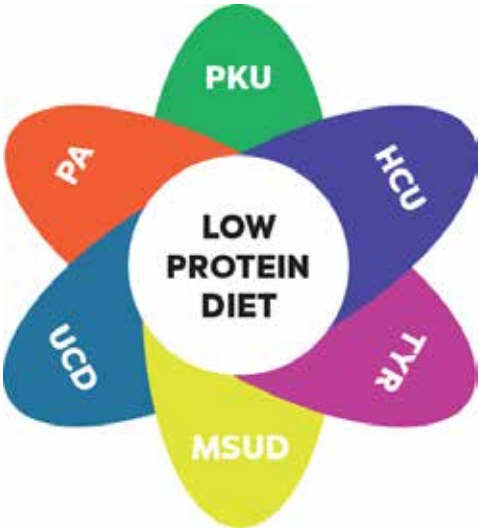
– Bridgett, a mother of a 6-year-old with HCU, who has tracked the low protein diet for her son's entire lifetime.

HOWMUCHPHE.ORG

HowMuchPhe.org is a comprehensive diet management system for patients requiring a low protein diet either measured in unrounded protein or exchanges for more than 7,000 foods.

The How Much Phe website allows you to search their data base, find related values, calculate your own recipes and create a daily food log. In addition, your subscription comes with responsive customer service.

While this website is designed for patients with PKU, it's a valuable resource because of the partnerships they have developed with many food companies to obtain unrounded protein content, which otherwise is hard to find.



METABOLIC DIET APP FOR HCY

Metabolic Diet App for HCY (homocystinuria, also known as HCU) is a website designed for patients with HCY. The website was designed with protein and methionine data provided by the Genetic Metabolic Dietitians International (GMDI) group to ensure accuracy and accessibility. In addition to the accurate data which can be used to track your daily nutrient intake, you can add your own foods and recipes, and export your diet records.

To find out more about Metabolic Diet App for HCY or to create an account, visitwww.metabolicdietapp.org/HCY.html.

DIET LOG

Date	Time	Food	Serving	Measurement	Protein

PART 6: MEDICATION, LABS AND DOCTORS

MEDICATION LIST

Name of Medication	Dosage Frequency (i.e. 2x per day)	Prescribing Doctor

LAB RESULTS

Lab	Date	Result

DOCTOR'S INFORMATION

Geneticist:		Clinic Affiliation:	
Address:		City, State, ZIP:	
Phone:		Email:	
Dietitian:		Clinic Affiliation:	
Address:		City, State, ZIP:	
Phone:		Email:	
Primary Care Physician:		Clinic Affiliation:	
Address:		City, State, ZIP:	
Phone:		Email:	

NOTES

RESOURCE GUIDE

For more information, tips and hints to managing HCU, visit these websites.

HCU NETWORK AMERICA BROCHURE

https://hcunetworkamerica.org/hcu-network-america-brochure/

RESOURCE LIST

https://hcunetworkamerica.org/wp-content/uploads/2022/04/Resource-List.pdf

CARETAKERS GUIDE TO CLASSICAL HOMOCYSTINURIA

https://hcunetworkamerica.org/wp-content/uploads/2019/02/Caretaker_Guide_HCU.pdf

WHAT SHOULD A CLINIC VISIT LOOK LIKE

https://hcunetworkamerica.org/wp-content/uploads/2018/11/Clinic_Visit_Infographic.pdf

EDUCATORS GUIDE TO CLASSICAL HCU

https://hcunetworkamerica.org/wp-content/uploads/2019/01/HCU_Educators_Guide_v4.pdf

EMERGENCY PREPAREDNESS TOOLKIT FOR ALL HOMOCYSTINURIA

https://hcunetworkamerica.org/wp-content/uploads/2020/03/Final-2020-Emergency-Preparedness-Toolkit-all-HCU-disorders.pdf

PARENT HANDBOOK FOR SPECIAL EDUCATION SERVICES

https://hcunetworkamerica.org/wp-content/uploads/2020/12/Parent-Handbook-for-Special-Education-Services.pdf

HCU COMMUNITY COOKBOOK

https://hcunetworkamerica.org/hcu-community-cook-book/

PATIENT STORIES

https://hcunetworkamerica.org/patient-stories/
Youtube Channel https://www.youtube.com/channel/UCdYChYZ3uMTGIpM0rBkmyaw

HCU NETWORK AMERICA