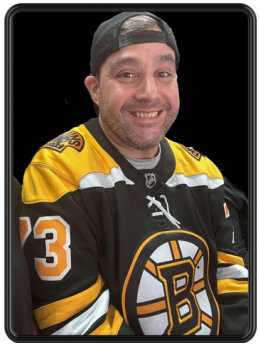
FAST TRACKING CLASSICAL HCU






A QUICK GUIDE TO LATE DIAGNOSED CLASSICAL HCU

HCU NETWORK AMERICA

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Thank you to HCU Network America's Patient-Parent Advisory Committee for helping develop the content of this booklet.

Disclaimer:
This booklet does not contain medical advice. Be sure to discuss any medical decisions with your doctor. All information shared by peers concerns their own personal experiences. The information contained has been scientifically reviewed and approved by the HCU Network America's medical advisory team.

INTRODUCTION

We are so glad you found us! If you are reading this, you have likely just been diagnosed with Classical Homocystinuria (HCU). As a recently diagnosed patient you may feel a bit of relief that you finally have a diagnosis, but likely also overwhelmed and isolated. You are not alone. We are here with you on this journey to support and assist you. We are patients and caregivers who want you to live your best life. This booklet will help give you a head start.

Take comfort in knowing that years of medical research have contributed to the establishment of a management plan for HCU, and that following the management plan can help you to maintain good metabolic control. Before you start to worry about medications, supplements, and diet, it is vital to establish care with your team of medical experts. Typically, an HCU patient needs to have, at a minimum, a geneticist and a dietitian, both of whom specialize in metabolic disorders. Not sure where to even start? Click here to see metabolic clinics throughout the U.S. You will likely need to travel a distance to visit an appropriate clinic, as homocystinuria is very rare, but it is worth it! Still, having trouble? We can help! Email us at info@hcunetworkamerica.org

Why is it so important to establish care with an experienced team? Two reasons: (1) majority of medical professionals are not familiar with HCU, nor do they know how to treat it; (2) not all patients with HCU are the same!

To put it quite simply, you are a rarity within a rarity. In the U.S., approximately 1 in 200,000 people have HCU. And of those with HCU, the treatment plans vary from person to person. What works for one patient might not for another. Some patients have a high protein or methionine tolerance, others low. Some take Betaine, some do not. Some are B6 (pyridoxine) responsive, others are not. Some count grams of protein, and others count milligrams of methionine. Therefore, it is vital that you connect with a specialist to establish a plan of care that is as unique as you.

While the plans may vary, the journey is one that you need not travel alone. Together, we are stronger. So start with the very first step: establish a plan of care with medical professionals.

Establishing Care

When establishing care, it is important to visit a metabolic center that specializes in HCU because they will help you set and reach your HCU goals. They can monitor the amount of homocysteine and methionine in your blood to find your protein tolerance, help you find the best medical food and formula that meets your needs, prescribe medications and supplements, and provide guidance.

To help you determine if a healthcare provider is right for you, we suggest asking the provider or their team the following questions and evaluating their answers, during your first appointment:

- How much time do you spend on an appointment?
- Who are the members of the HCU care team? (geneticist, nurse practitioner, dietitian, psychologist, social worker, etc.)
- What do you think are the important parts of HCU care?
- How much personal experience do you have with HCU?
- How open are you to new technologies, research, and therapies?
- Who is on call when you are not available?
- Are you available between scheduled appointments, and can I stay in contact with you remotely?

First Meeting

Preparing for the Appointment:

- Find out what you'll need before your appointment. Call your doctor's office and find out exactly what you need before getting there.
    - Ask if they want you to prepare a food log. If so, ask how many days they want you to record your food/beverage intake.
    - Do you need to have lab work done in advance? (They may go over this at the appointment)
    - Bring a list of your current supplements, medications, and prescribing doctors.
    - If able, understand the specifics of your insurance coverage. Does it cover laboratory studies, medical formula, low protein food, and medication? Your clinic should be able to help you with this. If not, email us! info@hcunetworkamerica.org

- Talk to your family members to collect your family health history, asking about medical conditions among your siblings, parents, aunts, uncles, cousins, grandparents, children and grandchildren. Don't forget to include your own health history.
- Write down questions and concerns for the doctor before your visit.

At the Appointment:

- Aim to get there early. Navigating a new hospital often requires extra time for parking and navigating the building.
- Bring everything you need, including your ID, insurance cards, and any other paperwork the doctor's office has requested.
- Your doctor needs accurate information to understand your health and make a diagnosis. Be open and honest about your health, lifestyle, and any challenges. Nothing is too big or small.
- Take notes during the appointment so you remember what is discussed.
- Bring a friend or family member to the appointment. They can be your support system before and after the appointment, and they can even take notes!
- Don't be afraid to ask questions, even if you think they might be embarrassing. Ask for clarification if you need help understanding something.
- Inevitably, you will have questions after your appointment. Find out how to contact your team if you have additional questions after you leave.
- Be sure to make the adjustments that your doctor has prescribed. It's easy to leave the doctor's office and forget about the changes you said you'd make.

Schedule Your Next Appointment

Whether with the same doctor or a new one, schedule your next genetics appointment ASAP so you don't forget. Schedule any other referrals or requested testing as well. You can always reschedule if needed, but get it on the calendar.

Initially, you will go to the clinic more frequently. This process allows your dietitian and physician to help you to establish an accurate protein tolerance. Once your levels stabilize and your team feels comfortable with them, they will see you less often.

Evaluate Your Visit

- Do you like your doctor?
- Do you feel more empowered about your care than when you walked in?
- Were your questions/concerns addressed?

These are really important questions to ask yourself. If the answer is "no", evaluate if there are things that could be done to make the relationship better.

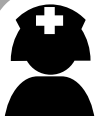
The first clinic appointment can be overwhelming. You might only meet a geneticist, but you might meet others. Don't worry, we have your back! Do know this could vary by clinic and how large the team is.

Geneticist

- Conducts physical exam
- Consults with patient on family and medical matters
- Confirms diagnosis
- Discusses family medical background

Dietitian

- Discusses diet
- Makes sure the patient is getting enough nutrients
- Answers food related questions



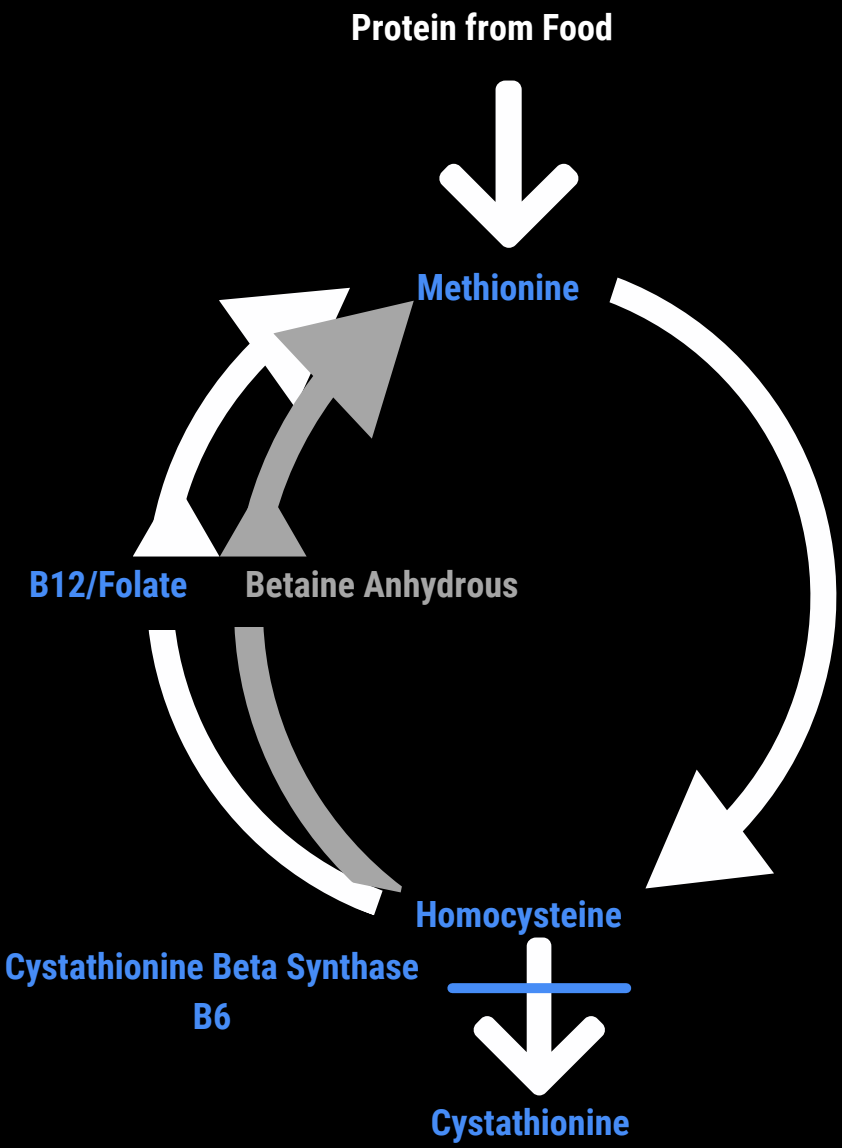
Nurse

- May organize educational groups and training
- Reviews lab tests
- May perform some duties of geneticist

Social Worker

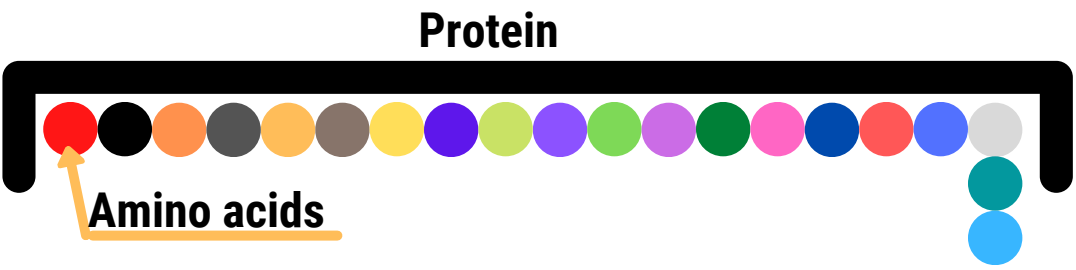
- Helps with access to low protein foods, formula, and medications
- Assist with various stages of diet management
- Assist with school-related diet resources

The Basics: What is HCU?



Words To Know

- **Amino Acids** are the building blocks of protein.
- **Enzymes** help the chemical reactions that break down food and assist with bodily functions at the cellular level.
- **Methionine (MET)** is one of the amino acids/building blocks that makes up protein.
- **Homocysteine (HCY)** is an amino acid that is typically converted to cysteine in the body, but builds up in the bloodstream in enzyme-deficient HCU patients.
- **Protein** is a macronutrient essential for building muscle and tissue in the body, and can be utilized as an energy reserve.



Classical Homocystinuria (HCU) is a genetic metabolic disorder where your body cannot metabolize (break down) the amino acid called Homocysteine (HCY) (hō-mō-ˈsi-stə-ˌēn) due to the deficiency of an enzyme called Cystathionine Beta Synthase (CBS). Individuals who cannot break down HCY have it accumulate and this also results in elevations in the amino acid Methionine (MET), which is converted to make HCY. MET and HCY are two of the building blocks of protein, which is the long-lasting energy source found in food and used to make more proteins.

MET is contained in proteins that we consume. Normally MET breaks down into the amino acid HCY which is then converted to cysteine. In classical HCU, the conversion to cysteine is defective and the byproduct HCY builds up and has very unhealthy and dangerous clinical effects. High HCY levels are harmful to the eyes, skeletal, vascular and central nervous systems.

How does this affect me?
If your enzymes can't break down HCY completely, it collects in your urine and blood and can cause a variety of symptoms. Most common complications include dislocations of the lens of the eyes, abnormal blood clots, osteoporosis, cognitive deficits, chest deformities, and/ or scoliosis. CBS enzyme activity varies among all patients, so different people experience different effects.

Should I feel different?
Not necessarily. The vast majority of people may feel fine; there are very complex processes in the body that may have negative effects in the long term. You are fine until you're not. Some patients have noted though that they feel "off" or experience brain fog when their homocysteine levels are elevated.

IMPORTANT Warning for Women!
Women who wish to start families should be very careful to follow the diet rules before, during, and while pregnant. This is very important for the health and safety of the mother while pregnant.

Key Warning Signs and Symptoms Include:

Patients are born with HCU. However, signs and symptoms may develop as early as infancy or adulthood. The symptoms with classical HCU vary greatly, with some affected individuals having only mild signs of the disorder; others may have many different symptoms, including some potentially life-threatening complications. The most commonly affected areas are the brain, the eyes, the skeleton, and the vascular system. Due to the symptoms and signs being nonspecific, this often leads to misdiagnosis and late diagnosis.

It is important to note that early treatment can prevent the development of symptoms listed on the next page or prevent further complications and worsening of already present symptoms.

Speak with your doctor about these potential symptoms and take this HCU Disease Checklist to your appointment.

The eyes

- Severe and progressive nearsightedness (myopia) at a young age
- Lens dislocation

The brain

- Developmental delays
- Cognitive deficit
- Clumsiness
- Psychiatric disorders: anxiety, depression, obsessive-compulsive disorder
- Behavior problems
- Seizures

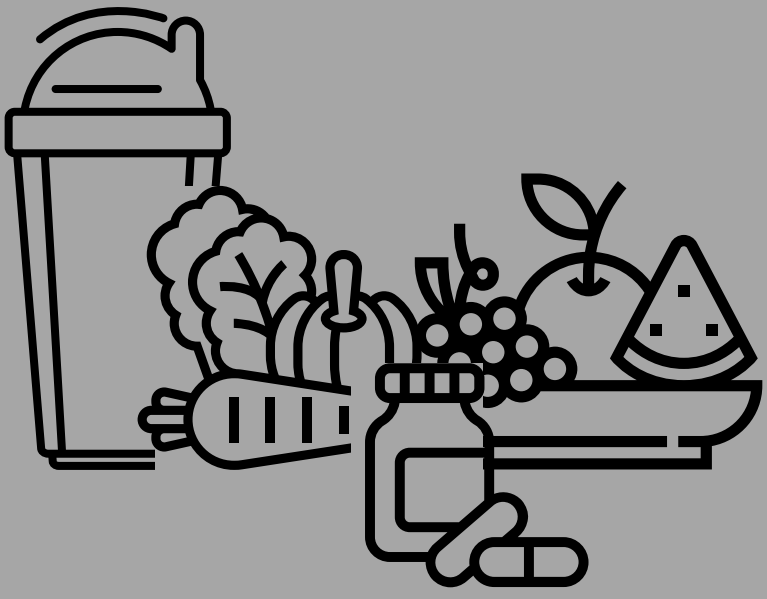
The skeleton

- Marfanoid features (specifically long limbs)
- Excessive growth and thinning or lengthening of the long bones
- Premature osteoporosis
- Scoliosis
- Restricted or excessive joint mobility in the hands
- Abnormally protruding or sunken chest
- Knocked knees
- Highly arched feet

The vascular system

- Blood clots, especially Deep Venous Thrombosis (DVT)
- Strokes (especially sagittal sinus thrombosis)
- Pulmonary embolism
- Mildly affected individuals may present as adults with blood clots as only problem

The Diet



Challenge:
The diet and formula are extremely overwhelming.

Solution:
Every clinic has its own method of folding you into the low protein diet. What has worked for many patients is the following approach:

1. Find a formula you like and get in a routine of taking all of it regularly
2. Eliminate meat – even a bite can mean a lot of protein!
3. Wean yourself off of high protein dairy products: milk, yogurt, cheese – explore low protein alternatives
4. Start supplementing pastas, breads, rice and flours with low protein alternatives. You may find gluten free alternatives at your store that are also lower in protein.

Challenge:
I'm so busy, I don't have the time!

Solution:
Try to plan your meals and meal prep! The biggest issue we have when we are busy is we get pressed for time, don't leave enough time to prepare something and then end up eating something that we know we really shouldn't be eating. Check out the meal plans and recipes in our HCU Community Cook Book page of our website for some inspiration.

Challenge:
I'm scared I'm going to gain weight.

Solution:
No one wants to unexpectedly gain 15 pounds. Starting the diet isn't just complicated because of the diet itself, but it can also be frustrating because some people experience unexpected weight gain.

First off, just because something is super low in protein, doesn't mean that it is equally low in calories and that you should binge eat it or even go for seconds. Taking your formula with each meal will help keep you full and from overeating. Try to incorporate high-fiber foods with each meal and if you are still hungry, go for more vegetables or fruit.

Don't forget that healthy weekly exercise can keep unwanted weight gain at bay as well.

Make Friends With Your Formula and Medication

You Want Me to Take What?
You may find that patients in the HCU community call formula by different names. Most commonly they will refer to it as “formula,” “milk,” or “protein shakes.” Whatever you choose to call it, the goals are the same. Protein is a necessary part of anyone’s diet, and for people with HCU it just happens to come in a different form: a methionine-free synthetic protein, so you can get the benefit of protein without taking in more MET which would only increase your HCY levels. Your protein supplement is an essential part of your diet, as the amount of protein you are allowed from solid food is likely quite restricted. Without your supplement, your total protein needs are far too low.

Everyone needs to eat every few hours to maintain energy, so it makes sense to have your formula when you eat. Much like food, formula works best when consumed over the course of the day, and not all at once.

Note: Following a low-protein diet but not taking formula can leave you with severe nutritional deficiencies and feeling weak. This is NEVER recommended.

How Do I Get My Formula?
Talk to your HCU dietician about your health and current lifestyle. Your dietitian should be able to send you samples of different types of formula that fit both your nutritional and personal needs. Pick one you like and stick with it. For some people, they will want a variety depending on circumstances and to avoid appetite fatigue.

Types of Medical Foods
The formula comes in a variety of formats. It can come as a powder in a can but also in single-serving packets, ready-to-drink pouches, and even in pill form! It comes in different flavors as well. Lots of people also choose unflavored formula and add flavoring.

Suggested Flavorings

Kool Aid, Tang, lemonade mix, Mio liquid water enhancer, iced tea, iced coffee, measured amounts of rice, coconut, or almond milk and juice. There are many types of flavorings on the market today. How you make your formula each day may vary, but be consistent in your daily intake.

- Mix exactly how much you need for each meal.
- Mix enough for the whole day and keep it cold in the refrigerator or cooler. Most types can be made ahead of time. Before drinking, be sure to give it a good shake.
- Keep an extra packet and bottle of water in your backpack, briefcase, or purse.
- Most types of formula cannot be heated up, because they will break down.

Tips for Taking Your Medication

It's very important that you take the medications your medical care team has prescribed for you.

- Use a pill box and fill it once a week to make it more convenient.
- Set an alarm on your phone to remind you to take your medications.
- Mix Betaine Anhydrous with a flavoring that you will like such as Sunny D or different juices.

What to Eat

People with HCU who are prescribed a low protein diet must avoid foods that are high in protein, such as meat, fish, poultry, dairy, soy, legumes (dried beans), or nuts. Some fruits and vegetables are higher in protein than others. MET is in almost everything except sugar, salt, oil, and water. Thankfully, many fruits and vegetables are naturally low in methionine/protein.

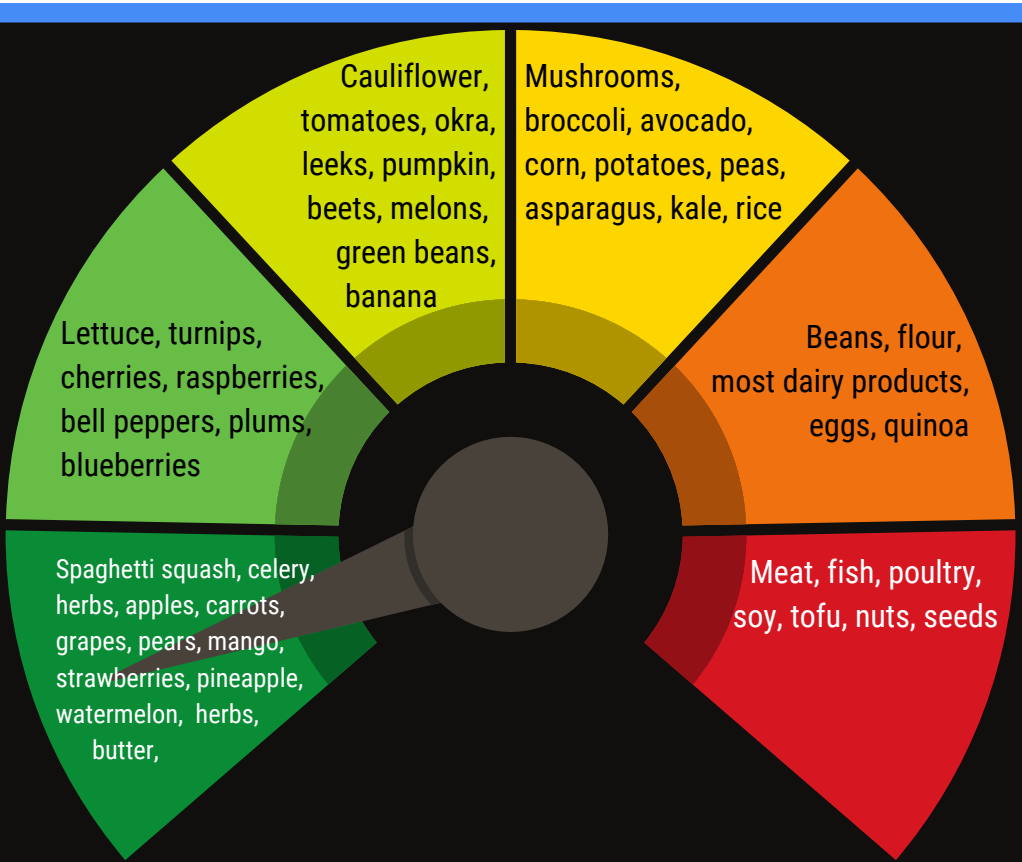
Check out the graphic on the next page to get an idea of some foods that are within your total protein tolerance. Always be mindful of keeping track of your intake so that you stay within your daily total protein limits.

What If I Miss High Protein Foods?

You can also order foods modified to be low in protein online or even in some grocery stores. See page 30 for a list of suppliers. These suppliers and most food stores can provide you with a variety of foods lower in protein, such as bread, baking mixes, pasta, sauces, desserts, pancakes, waffles, muffins, peanut butter, and even cereals and chocolates! You can even buy mixes for burgers, nuggets, and hot dogs. They may be more expensive but provide much more variety. Some states and insurance companies offer help paying for low-protein foods.

Making Good Food Choices

Many things have changed over the past few decades in the world of HCU. Thank you to vegans, vegetarians, and the gluten-free and dairy-free communities, we now have more options in grocery stores. You can often buy coconut milk yogurt, dairy-free cheese, dairy-free yogurt, gluten-free bread, and cassava pasta in a normal grocery store. Some, but not all, gluten-free, dairy-free, vegan, and vegetarian products are low in protein. Be sure to check the nutrition labels before you buy.



Foods shown in the green sections are generally lower in protein per serving. These are great foods to have readily available in case you're hungry and need something low to fill up on.

Foods in yellow tend to have a higher amount of protein in them and should be carefully measured and eaten in moderation. Foods in this section can vary greatly in grams of protein per serving so always be sure to weigh or measure your servings.

Foods in red should basically be avoided if possible.They are very high in protein and could cause your homocysteine level to rise if added to your daily food intake.

Note: The protein count will differ for foods when they're cooked, from when they are raw. Think about a baked potato vs potato chips. Brands can also vary.

Small Changes

It will probably be difficult to make huge changes in your lifestyle, but you can start with these lower protein options:

Higher Protein	Lower in Protein
• Bananas • Creamy soups • Ground beef • Mashed potatoes • Yogurt • Milk, half and half • Spaghetti • Desserts	• Apples, grapes, berries • Vegetable soup • Chopped and seasoned mushrooms or jackfruit • Mashed cauliflower • Coconut milk yogurt • Rice milk, non-dairy creamer • Bean thread noodles, rice noodles, zoodles • Coconut milk ice cream, sorbet, Oreos

How to Count Protein in Foods

Keep a diet record of EVERYTHING you eat and drink. Look at the example of a breakfast below. Don't be afraid to tell your clinic what you are really consuming. They are not there to judge you and will be able to help you better if they know where you are really starting from.

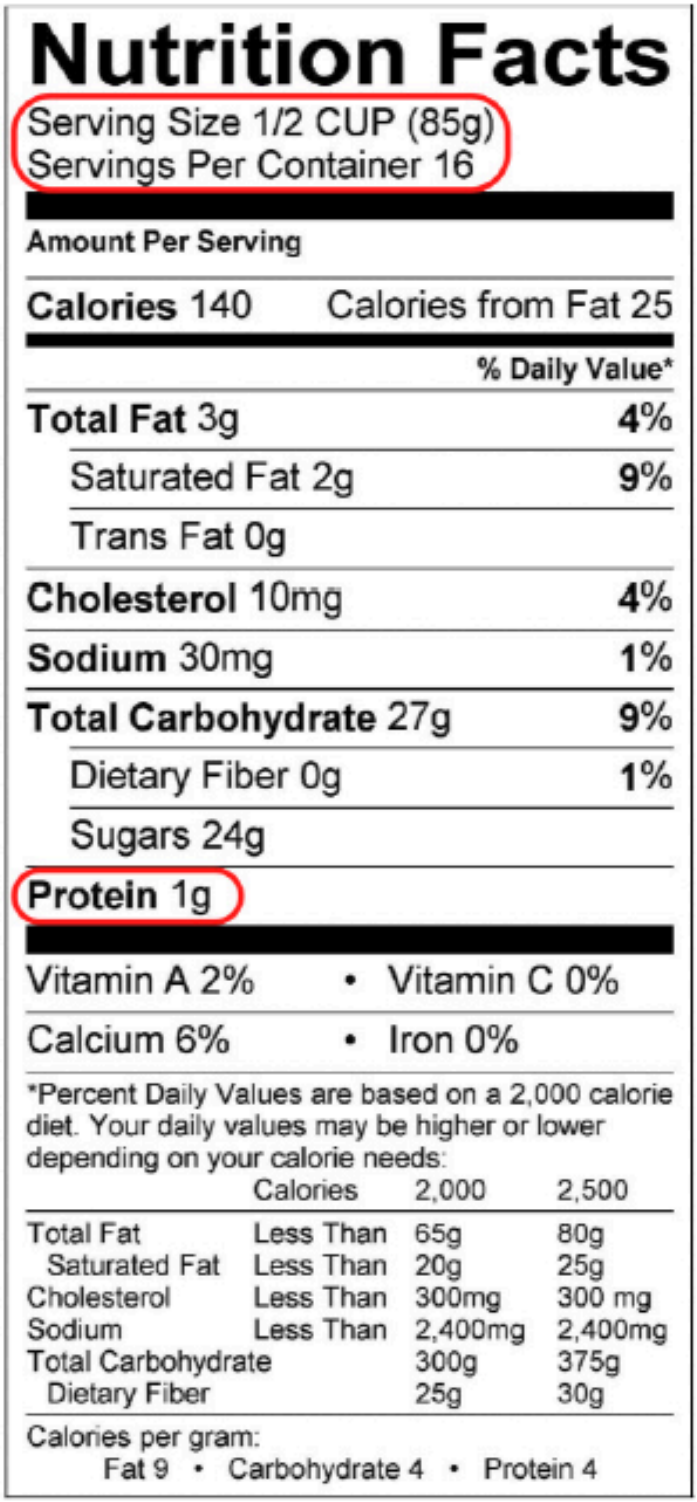
Monday - Breakfast

Quantity	Protein	Food Description
1 Sachet/pouch	0	HCU Express 15 formula powder mixed with lemonade
1 cup coffee	0.2 g	Coffee
1 tbs	0.2 g	Liquid Coffee Mate, original
1	0	Apple
2	2 g	Vans Gluten Free Waffles
2 tbs	0	Syrup
Total protein	**2.4 g**	

Packaged food will have a nutrition label. You can estimate how much protein is in these foods with a simple calculation.

It is important to not only look at the amount of protein listed on the nutritional facts but also to look at the serving size that equates to that amount of protein.

1. Check the serving size. This food is intended to be eaten in 1/2 cup servings.
2. Now look down to protein. This says 1 gram of protein for each ½ cup serving.
3. Multiply the number of servings you eat by the protein per serving to get grams of protein per serving. (number of servings you eat) X (protein) = (total protein)



Nutrition Facts

1 Serving Size 1/2 CUP (85g)
Servings Per Container 16

Amount Per Serving

Calories 140	Calories from Fat 25
	% Daily Value*
Total Fat 3g	**4%**
Saturated Fat 2g	**9%**
Trans Fat 0g	
Cholesterol 10mg	**4%**
Sodium 30mg	**1%**
Total Carbohydrate 27g	**9%**
Dietary Fiber 0g	**1%**
Sugars 24g	

2 **Protein** 1g

Vitamin A 2% • Vitamin C 0%

Calcium 6% • Iron 0%

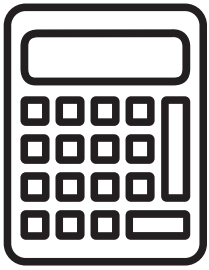
*Percent Daily Values are based on a 2,000 calorie diet. Your daily values may be higher or lower depending on your calorie needs:

	Calories	2,000	2,500
Total Fat	Less Than	65g	80g
Saturated Fat	Less Than	20g	25g
Cholesterol	Less Than	300mg	300 mg
Sodium	Less Than	2,400mg	2,400mg
Total Carbohydrate		300g	375g
Dietary Fiber		25g	30g

Calories per gram:
Fat 9 • Carbohydrate 4 • Protein 4

Other Options for Tracking Protein Intake and Helping Make Healthy Food Choices

Many people equate trying to follow a low-protein diet to losing weight. Like losing weight, some may opt for a paper diet log, but others opt for an app that helps them quickly find the exact protein content of foods and track it.



Things to Consider:

There are many options out there if you have decided to use an app to help you figure out the protein content and log what you are eating. The most important thing to consider is what is best suited for your lifestyle.

Reliable nutrition information: Is the nutrition information reliable? Can anyone upload and share nutrition information, or is it coming from a reliable source?

Rounded vs. Unrounded: Depending on your protein tolerance, you may be fine with accepting rounded protein – but if you have a lower tolerance, those 2/10th of a point start to add up!

Ability to export your diet log electronically: Part of the great wonders of tracking apps is being able to export your diet log to your dietitian.

Barcode, no barcode? Some apps now let you scan a food item – this allows you to scan an item and be on your way – one known risk is sometimes the information is inaccurate and regularly rounded.

Build/modify recipes: Many apps let you build recipes and modify them as you see fit – this lets you experiment or tweak things to fit your daily allowance.

Create new entries: Can't find a food item that you regularly eat? Many apps let you add new food items to your entries.

Low Protein Tracking Apps

Many low-protein food and formula companies have developed apps to help track protein. Here are a few of our top-rated items.

flok

Better understand your own health, communicate with your care team, and have the opportunity to contribute to a patient-led research platform.

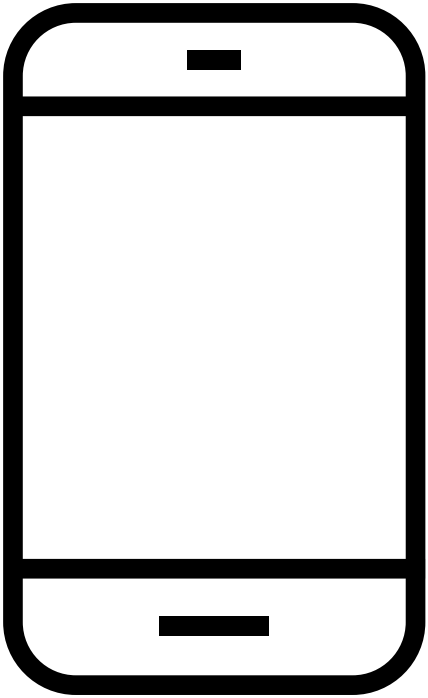
- App based
- Find the unrounded protein in over 10,000 items and growing
- Track unrounded protein, or exchanges.
- Add custom foods.
- Build/Modify recipes
- Meal plan ahead for a day, week or longer
- Export your diet log
- http://flok.org

HCY Metabolic Diet:

- Web based
- Syncs with data given to GMDI Metabolic Pro Database
- Daily nutrient tracker
- Add your own food or recipe
- Exports your diet records
- Allows you to track methionine and protein
- http://www.mdapp.org/

AccuGo for HCU:

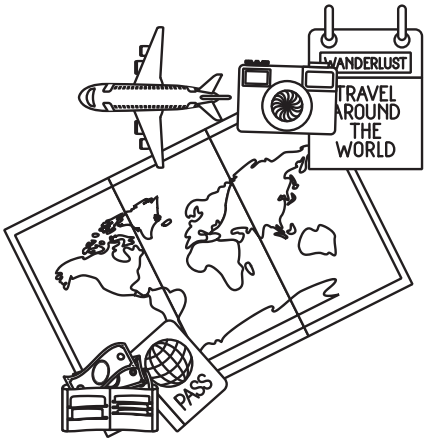
- Only available for iPhone
- Can work offline
- Can track protein or methionine
- Copy/move and delete entries with ease
- Allows you to menu plan
- Information based upon USDA based food items
- Ability to export daily food records
- https://apps.apple.com/us/app/accugo-for-hcu/id955040363



On the Go with HCU

Having to go out to eat can present a challenge to patients with HCU. It can be tough to access nutritional information and serving sizes, making counting grams of protein/milligrams of methionine feel more like picking lottery numbers than an attempt at science.

When possible, try to limit eating foods with high levels of protein. Instead, opt for vegetables, fruits, salads and other foods that you know are naturally low in protein. Lastly, know your staples. Choose a few foods that are usually available, and try to find out the exact protein counts.



Low Protein Restaurants: There is an App for That!

It's inevitable that at some point your friends will ask you to dinner, or you will be on vacation and need to grab some items to get you by. For this reason, we recommend a one of a kind app called Go Low Pro.

- Focused on you: Uses location to find locations near you. Filter by restaurant/store/brand to easily find what you need.
- Designed for low-pro: Gluten free is great, vegetarian is fine, but this app is designed JUST for low-protein diets to manage metabolic disorders.
- Brands you know: seeded with products by the members of the PartnershipforPKU.org, the app will help you find products your low-protein diet requires.
- Powered by you: Share new restaurants and stores complete with product details and pictures. The more you use the app, the better it gets for everyone!
- Traveling? Save room in your luggage: search by postal code and find locations near your destination to pick up low-pro supplies.
- https://golowpro.org/

Use Your Support System

Reach Out

Reach out to others who were diagnosed as adults. A little support goes a long way. With social networking websites and mobile phone apps, it is getting easier to meet other people like yourself. On Facebook it is as easy as searching for the word "HCU", or "Homocystinuria" in the search bar at the top of the page and saying hello to people who show up, or join some of the many HCU groups listed. Many states and regions also have official PKU and Allied Disorder support groups. Your clinic may be able to help you find them. Ask your clinic to put you in touch with other people with HCU and keep you informed about upcoming events. HCU Network America has a list of support groups on its website. Be sure to subscribe to HCU Network America's newsletter, the HCU Herald, to stay up-to-date on events and new resources.

Reach out to HCU Network America to join one of its Virtual Meetups, or be connected with a mentor!

Create Support

Support is so important. Your family and close friends, peers, or co-workers can provide a source of support. You don't need to tell everyone but try to find a few people you can turn to when you need a listening ear or who can learn some basics and help you.

For those you invite to this journey, set clear boundaries on how they can help.

DO

- DO ask me questions about HCU when you are curious.
- DO treat me the same way you treat your other friends.
- DO respect my feelings about HCU.

DON'T

- DON'T ask me "should you be eating that?"
- DON'T ask me if I'd like "just a bite" of something you know I can't eat
- DON'T look at/comment negatively on my diet.
- DON'T comment on how bad my food or formula may smell.

Stay Motivated, Not "Perfect"

Many people with HCU feel like they should always have blood levels of homocysteine readings in their target range, and become frustrated when, despite their best efforts, they don't reach their goals. This doesn't have to be a source of stress if you accept the fact that good metabolic care doesn't mean being perfect. Sometimes the elevated level is out of our control, as can happen if you are coming down with a cold. If you forgive yourself for the occasional higher level, you'll be relieved of the stress associated with trying to achieve perfection. You'll likely reap more rewards from this approach in the long term.

A Diagnosis Impacts More Than You!

Depending on how much you've shared with your friends and family about your diagnosis, these changes may come as a surprise. Just like you, they may find it difficult to process and struggle to understand the importance of the diet and what it really entails. It's not that they don't want you to be healthy, but they may struggle even more than you to understand your new needs.

Example:
Your family is celebrating by going out to eat; they pick an upscale steakhouse and ask if that is okay with you. Before, you wouldn't think twice about ordering a steak with its accompanying sides; now, your only choice is to get a salad and baked potato. You convey to them that your options are limited and that you'd prefer to go somewhere else. They get upset, leaving you feeling left out and making you feel like they don't understand and aren't being supportive. Try to remember that this is an adjustment for both sides, and it will take some time to find a compromise and routine in what is otherwise a standard scenario.

These situations will make developing a support network that much more important to your success!

My journey to diet was different than most. For more than 50 years, I was off diet due to a misdiagnosis of Marfan syndrome. When I was finally diagnosed with classical HCU at the age of 54, I knew I would have to make some changes to my diet to stay alive.

The Early Years

I was born in 1957, before the advent of newborn screening. I was the second of four children born to my mom and dad. I was an easy baby, at least according to what I have heard. I met my developmental milestones, such as crawling, standing, walking, and talking, with no delays. It wasn't until I was 2 years old that my parents noticed that I was having trouble with my eyesight. After an initial visit to an optometrist who discovered I had dislocated lenses, I was sent to an ophthalmologist for further evaluation. Because dislocated lenses do not occur in very many conditions and no one yet knew about HCU, I was diagnosed with Marfan syndrome. Throughout elementary school, I needed to sit in the front row in class, and my parents would always need to speak to the teachers to let them know about my vision difficulties.

Junior High School, High School and College

In junior high school, I started wearing contact lenses, which improved my eyesight tremendously. Although my eyesight at this time was improving, a routine physical exam revealed I had scoliosis. I tried physical therapy for several months, but in the end, I needed to wear a back brace for 2 1/2 years.

During this time, and later in high school, I was pretty much a typical teen. In high school, we had an open campus, which meant we were able to leave the school at lunchtime, and about a block away were all kinds of tempting lunch offerings. I often ate pizza, chili cheeseburgers, fries, milkshakes, ice cream, and more. I'd spend the night at friends' houses, go to movies, eat popcorn and candy, and never look back.

I grew up in a family where food was always at the center of everything. Thanksgivings were an elaborate affair, with an abundance of appetizers, turkey, mashed potatoes, sweet potatoes, stuffing, and more desserts than you could imagine. My cousins and I would eat and eat, and the next day, we would attack the leftovers again. Christmases were the same. We always spent time with our aunts and uncles, feasting on homemade tamales, rice, and beans.

I'd spend my summers at Lake Tahoe with my grandparents, who would take me out to fabulous restaurants about once a week on my grandfather's day off. I was exposed to a lot of different things and developed a taste for oysters on the half shell, tournedos of beef, escargot, and vichyssoise! My birthday was in mid-July, and my grandfather would almost always surprise me with a black forest cake for my special day.

During college, it was more of the same. I'd go on dates before my sorority dances, eating steak, prime rib, pasta, creamy soups, and, of course, cheesecake or crème brûlée for dessert! On lazy weekends, I would devour bagels and cream cheese, and omelets stuffed with cheese, sausage, or bacon.

The Later Years

After college, I married, adopted two children, and began my career as a special education teacher. In my late 20s and again in my 30s, I noticed that my eyesight was diminishing. One morning, while getting ready for work, I noticed it was like a curtain was being drawn down over my left eye. By the end of the day that eye was completely blind. It turned out that I had developed a retinal detachment, and I needed emergency surgery to repair it. At the same time, my doctor also removed my lens in that eye. I would again have surgery on my right eye in my 40s to remove my lens in that eye and repair a second retinal detachment. This time the surgery was much more difficult and as a result, I ended up with very little vision in that eye.

At the age of 46, I suffered a stroke. It began with typical stroke symptoms, such as trouble walking, confusion, and difficulty speaking. I spent four days in the ICU while doctors ran many tests, including MRIs of my head, neck, and brain. Luckily, the stroke was mild, and I recovered nearly all my functionality, but I was left with my right internal carotid artery being 100 percent occluded.

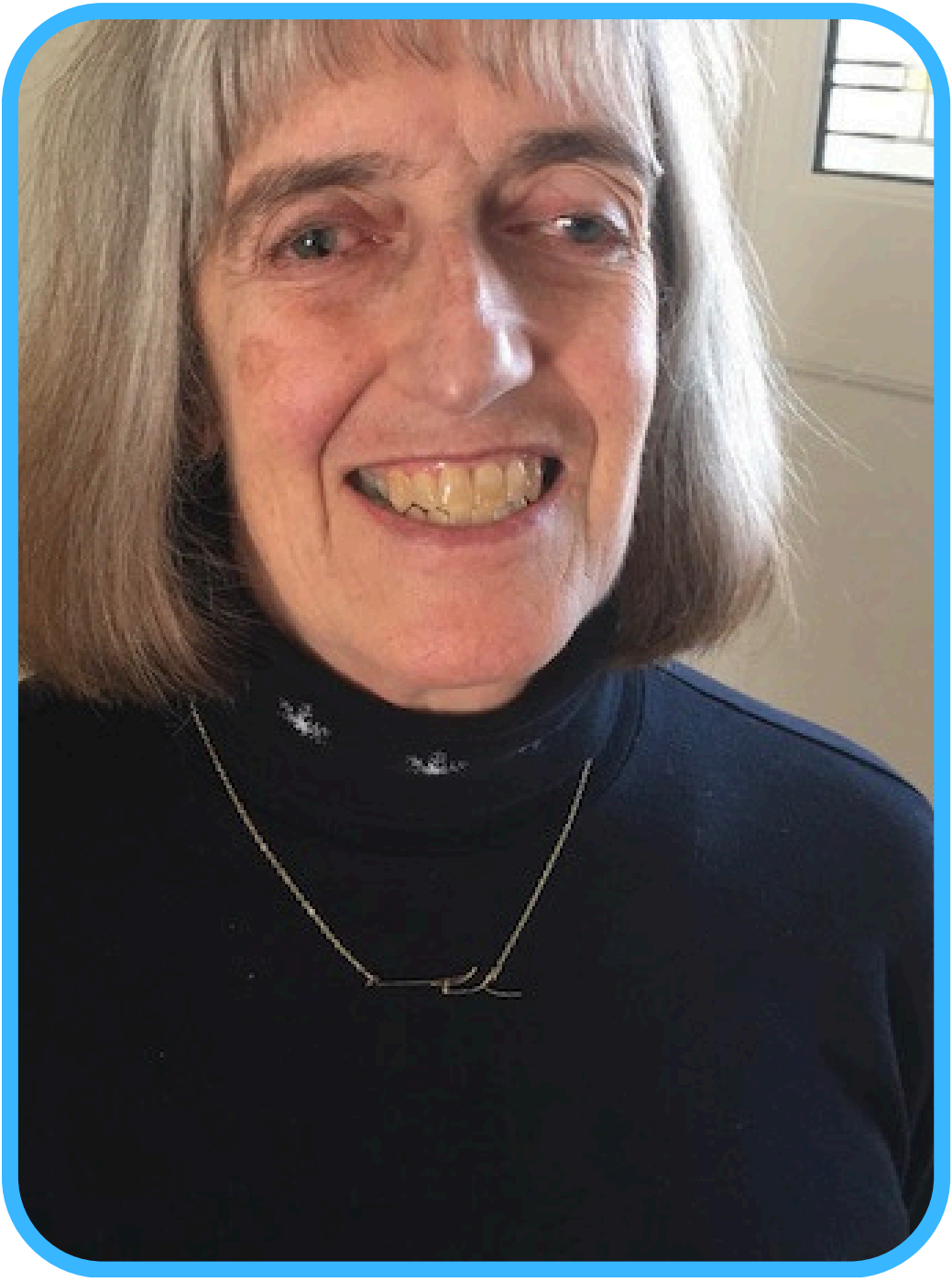
As if this wasn't enough, in my early 50s, I was hospitalized twice with blood clots in my lungs – four clots the first time and two more on the second occasion. When I was released from the hospital, I started doing research online. I had learned a lot about Marfan syndrome, but nothing I had read ever mentioned strokes or blood clots. While looking at the Marfan website one day, I noticed something called homocystinuria under "related disorders." I started reading about its symptoms –

dislocated lenses, scoliosis, vascular issues, strokes, and blood clots – and became very intrigued. Could it be that I had this disorder instead? I had been through the Marfan clinic at Stanford Hospital, and they, too, thought I had Marfan syndrome. I called the clinic and told them what I had read and that I thought I might have homocystinuria instead. They told me what blood tests to have done and to report my numbers back to them. My homocystine levels should have been between 5 and 15, but they were 412! I was told that I needed to be seen by one of Stanford's geneticists as soon as possible. At my appointment, the doctor ran a genetic test and a short time later I had my answer. I was diagnosed with classical homocystinuria In April 2011. Since I was found to be nonresponsive to B vitamins, I quickly had to start a strict low protein diet for life.

I remember how difficult it was at first to wrap my head around the fact that I would have to follow a stricter than vegan diet for the rest of my life. Because the dietitians at my clinic were more concerned about helping the parents of newly diagnosed pediatric patients, I was largely left to figure out the diet on my own. The dietitians gave me some PKU cookbooks and a book listing the protein content of many foods. There were supplemental materials in these books which explained how to measure my food on gram scales and track my daily intake of protein. After beginning there, I found other websites online that were specifically designed for diets like mine. One of my favorites has been Cook for Love which has hundreds of really tasty low protein meals and treats. As I grew more confident in managing my diet I began developing some of my own low protein recipes. I am very thankful that my mother was a wonderful cook who taught me a lot about cooking. Because of her guidance and training, I have never been afraid to try to create new dishes.

A Happy Ending

It has been over 13 years since I was diagnosed, and I am happy to say that I am doing very well. My levels have stabilized and my homocysteine levels are generally under 25. It is not always easy to manage my low-protein diet, especially at holidays and family gatherings, but I know it is something I need to do. I have had significant challenges throughout my life with my eyes, spine, and neurological systems because of being misdiagnosed and off diet for so long. Perhaps if I had been on diet and had received proper treatment from the beginning, I might not have had as much trouble with my health. But I am grateful to have done as well as I have.



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Low Protein Food Companies

- Lil's Dietary
  - 773-239-0355
  - https://lilsdietary.com/
- PKU Perspectives
  - 866-758-3663
  - https://www.pkuperspectives.com/
- Taste Connections
  - 310-413-6499
  - https://tasteconnections.com/

Food and Formula Companies

- Cambrooke
  - 866-456-9776
  - https://www.cambrooke.com/
- Zoia Pharma
  - 877-379-9760
  - https://www.zoiapharma.com/

Formula Companies

- Abbott Nutrition
  - 800-551-5838
  - https://abbottnutrition.com/infant-and-new-mother#metaboli
- Mead Johnson Nutrition
  - 800-457-3550
  - https://www.hcp.meadjohnson.com/products/metabolic-products/
- Nexus Patient Services
  - https://nexusmedicalnutrition.com/home
  - 833-875-0200
- Nutricia North America
  - 800-365-7354
  - https://www.nutriciametabolics.com/
- Vitaflo
  - 888-848-2356
  - https://www.vitaflousa.com/

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