

What should a clinic visit look like?

You may meet with:

The people who are helping treat you or your child's homocystinuria should be working as a team. They each play an integral part in the patients' health. Not all clinics will have each of the people listed below, but if they do, they each play a very important role. Understanding each person's role will help you advocate for your best care.



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
- May provide food related games



Nurse

- Tracks or screens for developmental progress
- May organize educational groups and training



Social Worker

- Helps with access to low protein foods, formula or other medication
- Assist with various stages of diet management
- Assist with school related diet resources



Clinical Psychologist

- Conducts social and cognitive developmental assessments
- ensures patient is socially and cognitively on track

What to bring:

- A list of your current medication and dosage
- Bring your diet log
- Be honest about how you are handling the diet,
 - They can't help you if they don't know you are having difficulties
 - A false diet log in conjunction with labs can lead to prescribed diet changes which could be hurtful!

They may perform:

- A height and weight check
- Measure the circumference of your head
- Routine labs, such as methionine, homocysteine, plasma amino acid panel, B12, Folate and Prealbumin
- Cognitive and social assessments
- Annual eye exams
- Checks spine for scoliosis
- Dexa Scans – every 3-5 years

What is a safe level for my homocysteine?

ANSWER: it depends on the disorder you have. We know that risk for thrombosis increases with total homocysteine greater than 120-150 mcmol/L. For classical CBS deficiency the Morris et al guidelines recommend below 80-100 mcmol/L. Some doctors recommend less than 80 mcmol/L and some as low as less than 30 mcmol/L to be safe. For B6-reponsive, most doctors recommend less than 50 mcmol/L.

How often do need to get blood work?

ANSWER: it depends on your age. Infants and young infants need to be checked often (initially every week, then every month or two). As one stops growing as fast this can be spread; children usually every 3 months. Older adolescents and adults on stable therapy with no major life issues (like surgeries), diet changes, or medication issues, many doctors check about every 4 to 6 months.

Will homocysteine levels change or increase if I or my child gets ill?

ANSWER: Usually it is yes. Total homocysteine increases during body stress, which is usually related to inflammation, thus everyone's homocysteine goes up with illness, and even more so in individuals like CBS patients who have difficulty processing homocysteine. Levels will gradually decline after illness.

If so, do we as caregivers adjust the diet to help counteract this?

ANSWER: Doctors usually encourage increased fluid and calories, but check with your metabolist and metabolic dietitian. It also helps make sure they know you are ill.

Where do you find low protein foods?

ANSWER: Talk to your metabolic dietitian for help. Different communities and areas have different resources. Other tricks include gluten free things. Metabolic food companies like Cambrooke have resources online. (To see a list of low protein food vendors, please visit <https://hcunetworkamerica.org/formula-and-medical-food-vendors/>)

If I stay on diet will I avoid possible symptoms?

ANSWER: That is the goal, if you have a disorder which responds to diet - Classical HCU patients need the diet. In classical CBS, we know that diet usually coupled with medications (like betaine) leading to homocysteine less than 100 mcmol/L in the first 4 years of life, leads to individuals having normal intellect and growth. You may though have symptoms if, e.g. you need emergency surgery.

If I already have symptoms, can they be reversed by lowering my levels?

ANSWER: Sometimes. In individuals with CBS deficiency whose eye lenses have started but not completely dislocated, lowering homocysteine (rapidly) may be able to prevent the lens dislocation. Abnormal growth can be normalized, if individual is still growing. If some intellectual loss, you can stabilize, but not reverse if damage is already done. It's possible bone loss can be reversed. But it is critical to control levels to prevent further problems.

As my child gets older will they be able to increase the amount of protein, they can eat daily?

ANSWER: As we get older, we get heavier. Most children's total amount of protein they can eat increases as they get older (since usually they get heavier). Ironically, even though the total increases the amount per kg weight may actually decrease as we become adults.

Why do we avoid foods high in methionine, if we are more concerned about homocysteine levels?

ANSWER: Methionine is an essential amino acid (not made from other amino acids) which we get from protein in food. We convert it to homocysteine so by lowering intake we make less homocysteine.

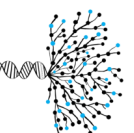
Hydration is very important to people with HCU – is it even more so when playing sports or any type of physical activity?

ANSWER: Yes, hydration is very important. We know that risk for blood clots goes up (for everyone, but more so if have HCU) when we are dehydrated. If you already have a risk for blood clots, like those with HCU, the recommendation is not to become dehydrated ever.

Why do I need to drink my dietary formula in combination with a low protein diet?

ANSWER: Despite being on a low methionine diet, your body still needs protein. Your formula provides you with the proper type of protein (i.e. amino acids other than methionine) along with vitamins and other important nutritional elements you lack with a low protein (methionine) diet. Without the protein in your formula, your body will break down your muscles causing your methionine levels to increase and then as a result homocysteine to rise. In addition, because your formula provides proper protein and nutrients, it allows your body to feel full and satisfied which helps you follow the low protein diet.

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