Metabolic.ie
National Centre for Inherited Metabolic Disorders

Information for Pharmacists

What is HCU?

Homocystinuria (HCU) is an inherited metabolic disorder affecting the metabolism of foods containing protein. People with HCU cannot use protein in the normal way. Protein is made up of amino acids, one of which is methionine.

In HCU, the enzyme cystathionine beta synthase (CBS) which breaks down methionine and its byproducts is absent or only present in small amounts. This causes a build up of methionine and homocystine. This build up can cause problems in several parts of the body including the central nervous system, eyes, skeleton and blood systems unless treatment is commenced. Some methionine is essential for growth. HCU is diagnosed by newborn screening in Ireland.

HCU can be classified into two forms:
• pyridoxine (vitamin B6) responsive HCU
• pyridoxine (vitamin B6) non-responsive HCU

If a patient is diagnosed with non-pyridoxine responsive HCU, treatment involves medications and changes to diet. HCU is treated by restricting the natural protein in the diet. The protein foods are replaced by a synthetic amino acid mix and the diet supplemented by a variety of low protein foods available on prescription. These low protein foods provide energy, bulk and variety in the diet. This is the more common type of HCU in Ireland and requires a **diet for life.**

Prescriptions

Children and adults with HCU will require regular prescriptions for their low protein foodstuffs. These items are an integral part of the treatment of HCU and without these foods the dietary management will not be successful. In Ireland, all patients

are entitled to the long term illness card which entitles them to order low protein foods and their prescribed synthetic protein/amino acid supplement without cost. In some cases a medical card may be issued.

Low protein foods: There are a large variety of low protein foods available from several manufacturers. The patient will often prefer one manufacturer to another for different items. It is very important to encourage variety in the diet so that patients will not resort to seeking restricted foods and thereby upsetting their dietary management. We provide an updated list of manufactured food each year.

Amino Acid Supplements: Every patient following a low methionine diet requires an amino acid supplement to replace the forbidden high protein foods. It is vital they have a constant supply of this amino acid supplement. If they do not receive this supplement then their methionine levels will rise because the patient will become catabolic. If this continues over time it will effect the patient's growth and development.

There are several types of amino acid supplement, prescribed according to age and suitability for the patient. Once the child or adult is established on one they tend not to change for several years. Supplies can therefore be ordered in advance to ensure a constant intake. There are a range of amino acid supplements for other metabolic disorders which have similar label colour, label design and names to the methionine free supplements. These are **not** suitable for the treatment of HCU.

What if you have supply issues?

Should you experience any problems obtaining any of the low protein foods or amino acid supplements it is worth contacting the manufacturer directly as they often have sufficient stocks of the products.

A metabolic dietitian can be contacted at any time during office hours to discuss any problems or concerns you may have about the prescriptions for the patients with HCU.

Trouble-Shooting

- When new products become available on prescription, they are often not easy to obtain because the wholesaler you use may not have obtained supplies. It is best to put in your order as usual and then contact the supplier and order the product direct until stocks are available locally.
- There are some low protein foods produced by small companies that may not be available from your wholesaler. If you have difficulty obtaining any of the low protein foods they should be ordered directly from the company.
- Many gluten free products are similar in appearance to the low protein foods, **but are not suitable** for the patient on a protein restricted diet as they often have protein added in the form of egg, milk or soya. This will increase the protein content of the product to a level too high for patients following a low phenylalanine diet. Many low protein products are Gluten free, but **Gluten free products are not necessarily low protein**

For more information about HCU contact:

NCIMD

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Nutricia	United Drug	01-2890283
Freephone number: 1800 923535		
Email: metabolic.ireland@nutricia.com		
PK Foods	All Phar	01-4041600
Email: ellis@glutenfree-foods.co.uk		
Juvela	United Drug	01-4632300
Freephone number: 1800 405090		01-4598877
Email: lowprotein@juvela.co.uk		
Promin	Biofact	01-612544
UK number: 0044161 474 7576		
Email: info@prominpku.com or katya.sheridan@firstplaydf.com		
Taranis	Biofact	01-612544
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