

Inborn Errors of Metabolism and Dietary Treatments

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Diet and IEM conditions

Over 30 conditions detected by NBS are treatable with medical nutrition therapy (diet) which includes special medical foods and formula.

Many more conditions that would benefit from early dietary treatment are not detectable by NBS.

Maple Syrup Urine Disease (MSUD)

Diet limited in: leucine (LEU), isoleucine (ILE), valine (VAL)

Formula needed: yes, also amino acid supplements

Special low-protein foods needed: yes



What happens in MSUD?

Without treatment, a baby with classic MSUD will experience lethargy, loss of appetite, feeding problems, convulsions, coma, respiratory distress, and death.

Even with treatment, each childhood illness puts the patient at risk of the above problems and prompt ER treatment is necessary when there is illness and vomiting. These risks continue into adulthood unless the patient gets a liver transplant which is considered curative.

Diet in a baby with MSUD

A special recipe would be made up daily:

- 60 grams of Ketonex-1 powder
- 183 mL of Similac infant formula, ready-to-feed type
- 6.1 mL of isoleucine solution
- 7.9 mL of valine solution
- 25 mL of Polycose liquid
- Add water to make a total of 19 oz (569 mL)

*the above recipe would change frequently depending on weekly blood levels of amino acids and also the baby's weight and appetite

Homocystinuria (HCU)

Diet limited in: methionine (MET)

Formula needed: yes

Special low-protein foods needed: yes



Special low-protein foods

Regular baked goods and rice would be too high in protein, so there are options.



What happens in homocystinuria?

Without proper diet, blood levels of homocystine rise and the patient is at risk for dislocated lens of the eye, blood clots, and stroke at an early age. Other possible problems include cognitive delay and bone problems.

If the diet is low protein (without enough formula used), the patient is at risk for protein deficiency: hair falling out, skin rashes, poor immunity, children won't grow

So low protein diet + formula = best outcome

Example diet, adult with HCU

10-12 grams of regular food protein (150-200 mg MET):

- 2 cereal bars
- spinach veggie wrap
- 6 cups lettuce
- 3 cups juice or punch
- 1 cup ranch dressing
- 2 cups of fruit

“SAFE” protein intake (MET-free) of 40-60 grams from formula:

- 1/3 to 1/2 can Hominex-2 (120 grams of safe protein per can)
- OR
- 3-4 HCU Coolers (15 grams of safe protein each)

Tyrosinemia (TYR)

Diet limited in: tyrosine (TYR) and phenylalanine (PHE)

Formula needed: yes

Special low-protein foods needed: yes



Propionic Acidemia (PA)

Diet limited in: VAL, ILE, MET, THR

Formula needed: yes

Special low-protein foods needed: sometimes



Methylmalonic Acidemia (MMA)

Diet limited in: VAL, ILE, MET, THR

Formula needed: yes

Special low-protein foods needed: sometimes



Glutaric Acidemia (GA-1)

Diet limited in: lysine (LYS) and tryptophan (TRP)
 Formula needed: yes, for younger children
 Special low-protein foods needed: sometimes



Urea Cycle Disorders (UCD's)

(there are several kinds, not all detected by NBS)

Diet limited in: overall protein
 Formula needed: usually for best outcome
 (give extra essential amino acids)
 Special low-protein foods needed: sometimes



Galactosemia

Diet limited in: galactose (and lactose)
 Formula needed: yes, soy usually for infants and young children
 and soy (or other non-dairy) milk later



Fatty Acid Oxidation Disorders

(There are many kinds of FAOD's)

VLCAD: this disorder is treated with a special oil in the diet
 (medium chain triglycerides or MCT):



Other Formulas for IEM Treatment

These are mainly used to provide calories in a low-protein diet.



Amino acids used to treat IEM's



Questions?

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