KENTUCKY NEWBORN SCREENING PROGRAM

PARENT TEACHING SHEET

Kentucky Newborn Screening Program, 275 East Main Street, Frankfort, KY 40621, Phone (502) 564-3756, Fax (502) 564-1510

2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBDH)

In order for the body to use protein from the foods we eat, it is broken down into smaller parts called amino acids. Special enzymes then make changes to the amino acids so the body can use them. 2MBDH occurs when an enzyme, called 2-methylbutyryl-CoA Dehydrogenase, is missing or not working properly.

IF NOT TREATED

Each child with 2MBDH deficiency may have somewhat different effects. Symptoms can be mild or absent to very severe. Most babies are healthy at birth. Symptoms can develop just a few days after birth or later in childhood. Symptoms can be triggered by prolonged periods without food, infections, or eating an increased amount of protein-rich foods.

TREATMENT OPTIONS

Some children may never develop symptoms and may never need treatment. Lifelong treatment may be needed for babies that are symptomatic. Treatment options include:

- Avoid going a long time without food: Some infants and young children need to eat frequently. How often your baby needs to eat will be determined by your metabolic specialist.
- Low-protein diet: A food plan low in protein is sometimes needed. Most food in the diet will be carbohydrates (bread, cereal, pasta, fruit, and vegetables.)
- Medical foods and formula: special low-protein flours, pasta and rice that are made especially for people with organic acid disorders.
- Medications: Some children benefit from taking a medication called L-Carnitine. This medication helps body cells make energy. It also helps the body get rid of harmful wastes. Your doctor will decide if your child needs L-Carnitine

IF TREATED

With prompt and careful treatment, children who have shown symptoms of 2MBDH have a good chance to live healthy lives with typical growth and development. Even with treatment, some children still have repeated bouts of metabolic crisis that require close monitoring throughout their life.