

# KENTUCKY NEWBORN SCREENING PROGRAM

## PARENT TEACHING SHEET

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### Malonic Acidemia (MAL)

#### CAUSE

Malonic Acidemia (MAL) is an inherited disorder in which the body is unable to process certain proteins and fats properly. The effects of methylmalonic acidemia, which usually appear in early infancy, vary from mild to life-threatening. MAL is characterized by an accumulation of methylmalonic acid in the blood stream, which leads to an abnormally low pH (high acidity) in nearly every cell in the body (metabolic acidosis) and a higher than normal accumulation of ketones in the blood stream (ketosis). If left untreated, metabolic acidosis is often fatal.

#### SYMPTOMS

- Hypoglycemia
- Lactic acidosis
- Marked lethargy
- Developmental delay
- Seizures
- Vomiting
- Failure to thrive
- Low muscle tone/strength
- Metabolic acidosis
- Deterioration of the function of the heart muscle
- Chronic kidney disease
- Inflammation of the pancreas

#### Treatment

Your baby will be on a restricted diet to avoid high fat foods. A dietician will help you plan the right diet to keep your baby healthy. Eating often will help keep your baby from having problems. Your doctor will prescribe L-carnitine supplements to help breakdown fats and remove harmful substances from the body. L-Carnitine is a natural substance. You will need a prescription from the doctor for L-Carnitine. Careful and prompt treatment helps children with MAL live the healthiest lives possible.

