

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

Ethylmalonic Encephalopathy (EE)

CAUSE

People with EE have problems breaking down fat and protein into energy for the body.

EE occurs when one of two different enzymes is either missing or not working properly. The job of these enzymes is to help make energy for the body by breaking down certain fats and proteins from the food we eat. They also break down fat and protein already stored in the body.

Energy from fat and protein keeps us going whenever our body runs low of its main source of energy, a type of sugar called glucose. Once glucose is used up, the body tries to use fat and protein with limited success.

IF NOT TREATED

EE can cause bouts of illness called metabolic crises. Some of the first symptoms of a metabolic crisis are:

- extreme sleepiness
- irritable mood
- muscle weakness
- Abnormal movements
- Blue discoloration in hands and feet
- Seizures
- poor appetite
- Chronic diarrhea
- Progressive developmental delay
- Rashes of tiny red spots (petechiae)
- It is very important to follow the doctor's instructions for testing and treatment.

TREATMENT OPTIONS

Your baby may need to be on a restricted diet in order to avoid the foods that his or her body cannot break down. A dietician or a nutritionist is someone your pediatrician may refer you to who can help plan a specialized diet for your baby.

Your baby's pediatrician may recommend L-carnitine, riboflavin, Q10s, and/or other vitamin supplements, which may help your baby produce energy and reduce damage to cells and tissues caused by a build-up of toxic substances.

IF TREATED

For some individuals with EE, detecting it early and beginning treatment may help delay some of the serious health outcomes associated with the condition.



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