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

Maple Syrup Urine Disease (MSUD)

CAUSE MSUD stands for "maple syrup urine disease". It is named for the sweet maple syrup smell of the urine in untreated babies. This condition is one type of amino acid disorder. People with MSUD have problems breaking down certain amino acids found in protein.

Classic MSUD, the most common form, is caused by the absence of a group of enzymes called "branched-chain ketoacid dehydrogenase" (BCKAD). The job of this enzyme group is to break down three different amino acids called leucine, isoleucine and valine. When they cannot be broken down, these amino acids build up in the blood and cause problems.

Symptoms start as soon as a baby is fed protein, usually shortly after birth. Some of the first symptoms are: poor appetite, weak suck, weight loss, high pitched cry, and urine that smells like maple syrup or burnt sugar.
Babies with MSUD have episodes of illness called metabolic crises. Some of the first symptoms of a metabolic crisis are: extreme sleepiness, sluggishness, irritable mood and vomiting.

IF NOT TREATED If not treated, other symptoms can follow: episodes where the muscles become tight and rigid and then go limp, swelling of the brain, seizures, high levels of acidic substances in the blood, called metabolic acidosis and coma, sometimes leading to death.

Symptoms of a metabolic crisis often happen: after going too long without food, during illness or infection and during stressful events such as surgery.

Without treatment, brain damage can occur. This can cause mental delays or spasticity. Some babies become blind. If not treated, most babies die within a few months.

TREATMENT OPTIONS There are a number of different forms of MSUD. The most common form, "classic MSUD", can be life-threatening and must be treated promptly to prevent serious health problems. Other forms are less severe. These milder forms are less common.

Lifelong treatment with the MSUD diet is necessary. Children are at risk for episodes of metabolic crisis when they don't follow the diet.

In addition to a low-protein diet, children are often given a special medical formula as a substitute for milk. This formula gives them the nutrients and protein they need while helping keep their BCAA levels in a safe range.

Your metabolic doctor and dietician will tell you what type of formula is best and how much to use. The diet is made up of foods that are very low in the BCAAs. This means your child will need to avoid foods such as cow's milk, regular formula, meat, fish, cheese and eggs. Regular flour, dried beans, nuts, and peanut butter also have BCAAs and must be avoided or strictly limited. Your metabolic doctor and dietician will decide on the best food plan for your child. The exact plan will depend on many things such as your child's age, weight, and general health. Your dietician will fine-tune the diet over time. Any diet changes should be made under the guidance of a dietician

IF TREATED With prompt and lifelong treatment, children with MSUD often have healthy lives with typical growth and development. Early treatment can help prevent brain damage and mental delays.

Even with treatment, some children still develop swelling of the brain or have episodes of metabolic crisis. Children who have repeated metabolic crises may develop permanent brain damage. This can cause lifelong learning problems, mental delays or spasticity.

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