

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

Methylmalonic Acidemia Mutase Deficiency (MMA mut def)

Cause MMA stands for "methylmalonic acidemia". It is one type of organic acid disorder. People with MMA have problems breaking down and using certain amino acids and fatty acids from the food they eat.

If Not Treated MMA causes episodes of illness called metabolic crises. Some of the first symptoms of a metabolic crisis are: poor appetite, vomiting, extreme sleepiness or lack of energy and low muscle tone (floppy muscles and joints).

Common blood and urine findings are: ketones in the urine, high levels of acidic substances in the blood, called metabolic acidosis, high blood ammonia levels, high blood and urine levels of glycine, high blood and urine levels of methylmalonic acid and propionic acid, high levels of other harmful substances, low platelets, low white blood cells and anemia.

If a metabolic crisis is not treated, a child with MMA can develop: breathing problems, seizures, stroke and coma, sometimes leading to death. A metabolic crisis can be triggered by: eating large amounts of protein, illness or infection, going too long without food and stressful events such as surgery. Without treatment, brain and nerve damage can occur. This can cause mental delays and problems with involuntary movements. Death is common in untreated babies and children.

Treatment Options Your baby's primary doctor will work with a metabolic doctor and a dietician to care for your child. Prompt treatment is needed to reduce the chance for mental delays and serious medical problems. Children with 'vitamin B12 responsive' MMA are given vitamin B12. In addition, most children need to be on a low-protein diet and drink a special medical formula. You should start the treatments as soon as you know your child has MMA.

Children who are having symptoms of a metabolic crisis should be treated in the hospital. During a metabolic crisis, your child may be given medications such as bicarbonate through an IV to help reduce the acid levels in the blood. Glucose is given by IV to prevent the breakdown of protein and fat stored in the body. Do not use any medication without checking with your doctor.

A food plan low in the amino acids leucine, valine, methionine, and threonine with limited amounts of protein is often recommended. Most food in the diet will be carbohydrates (bread, cereal, pasta, fruit, vegetables, etc.). Carbohydrates give the body many types of sugar that can be used as energy. Eating a diet high in carbohydrates and low in protein and fat can help prevent metabolic crises.

Your dietician can create a food plan that contains the right amount of protein, nutrients, and energy to keep your child healthy. It is likely your child will need to be on a special food plan throughout life.

If Treated Babies and children who have prompt and ongoing treatment may be able to live healthy lives with normal growth and development. In general, the earlier treatment is started, the better the outcome.

Children who respond to vitamin B12 treatment tend to do very well as long as treatment is continued. Children who are not treated until after they have symptoms may have lasting health and learning problems.

Even with treatment, some children develop life-long learning disabilities or mental delays. In addition, despite treatment, seizures, involuntary movement disorders, and kidney failure have occurred in some children.



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