PARENT FACT SHEET

DISORDER

3-methylcrotonyl CoA carboxylase deficiency (3MCC)

CAUSE

In order for the body to use protein from the food 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. 3MCC deficiency occurs when an enzyme is missing or not working properly.

IF NOT TREATED

Each child with 3MCC deficiency may have somewhat different effects. Babies with 3MCC deficiency are healthy at birth. If symptoms occur, they often start after 3 months of age. Some babies do not have symptoms until 6 months to 3 years of age. If not treated these babies could go into metabolic crisis which could lead to seizures, breathing problems, liver failure, coma or death.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist and dietician. Treatment is needed throughout the child's life.

- A food plan low in leucine with limited amounts of protein is sometimes needed. Most food in the diet will be carbohydrates (bread, cereal, pasta, fruit, vegetables).
- Medical foods and formula: special low-protein flours, pastas and rice that are made especially for people with organic acid disorders.
- Some children may benefit by taking a medication called L-carnitine.

IF TREATED

With prompt and careful treatment, children who have shown symptoms of 3MCC deficiency 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.

For more information go to the following website: http://www.newbornscreening.info