PARENT FACT SHEET

DISORDER

Carnitine Palmitoyl Transferase deficiency, Type I (CPT-I)

CAUSE

CPT-1 occurs when an enzyme is either missing or not working properly. This enzyme's job is to help change certain fats in the food we eat into energy. It also helps bread down fat already stored in the body.

IF NOT TREATED

CPT-1 deficiency can cause episodes of illness called metabolic crisis. Symptoms usually start between the ages of 8 and 18 months, although effects can occur earlier. Some of the first symptoms of metabolic crisis are: extreme sleepiness, behavior changes, irritable mood and poor appetite. Between episodes of metabolic crisis, people with CPT-1 deficiency are usually healthy. However, repeated episodes may cause brain damage that can result in learning problems or mental delays. Symptoms often happen after having nothing to eat for more than a few hours. Babies and children who are not treated can have: learning problems, delays in walking and other motor skills and liver, heart or kidney problems.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist and dietician. When necessary, treatment is needed throughout life.

- Infants and young children with CPT-1 need to eat frequently (generally every 4-6 hours) to prevent hypoglycemia or a metabolic crisis.
- A low-fat, low-protein, high-carbohydrate diet is often advised. A dietician will help you create a food plan that meets your child's needs.
- Some children and adults with CPT-1 are helped by taking daily L-carnitine or MCT oil supplements. Your metabolic specialist will prescribe these medications, if necessary.
- Contact your child's doctor immediately at the start of any illness.

IF TREATED

With prompt and careful treatment, children with CPT-1 deficiency often live healthy lives with typical growth and development. After 5 years of age, metabolic crises tend to happen less often and are not as severe.