



Newborn Screening FACT Sheet

Trifunctional Protein Deficiency (TFP)

What is TFP?

TFP is a type of fatty acid oxidation disorder. People with TFP deficiency can't break down fat into energy for the body.

What Causes TFP?

Enzymes help start chemical reactions in the body. TFP happens when a group of enzymes called "trifunctional protein" is missing or not working. TFP breaks down certain fats from the food we eat into energy. It also breaks down fat already stored in the body.

What Symptoms or Problems Occur with TFP?

[Symptoms are something out of the ordinary that a parent notices.]

Babies and children with early and childhood TFP have periods of illness called Metabolic Crises. Some of the first symptoms of a Metabolic Crisis are:

- too much sleepiness
- behavior changes (such as crying for no reason)
- irritable mood
- muscle weakness
- poor appetite

If a Metabolic Crisis is not treated, a child with TFP can develop:

- breathing problems
- seizures
- coma, sometimes leading to death

What is the Treatment for TFP?

The following treatments are often used for children with TFP deficiency:

1. Do not go a long time without food –

Babies and young children with TFP need to eat often to avoid low blood sugar and Metabolic Crises. They should not go without food for more than 4 to 6 hours. Some babies need to eat even more often. It is important that babies be fed during the night. Your dietitian can give you ideas for good night-time snacks. Dietitians know what are the right foods to eat.

2. Diet – Sometimes your child needs a diet low in fat (such as lean meat and low-fat dairy foods) and high in carbohydrates (such as bread, noodles, fruits, vegetables). People with TFP cannot use certain building blocks of fat called "long chain fatty acids." A dietitian can make a food plan low in these fats.

3. MCT oil and L-carnitine – MCT oil is often used for people with TFP. This special oil can be used in small amounts for energy. Sometimes the doctor will prescribe L-carnitine. This is safe and natural and helps body cells make energy. It also helps the body get rid of harmful wastes.

Do not use any medication without checking with your doctor.

Things to Remember

Always call your doctor when your child has any of the following:

- poor appetite
- low energy or too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- continued muscle pain or weakness
- reddish-brown color to the urine