

Fatty Acid Disorders Information for Parents

➤ Overview

Fatty acid disorders (also called fatty acid oxidation disorders) are a group of rare, inherited conditions that affect infants from birth. They are caused by enzymes that do not work properly. A number of enzymes are needed to break down fats in the body (a process called fatty acid oxidation). Problems with any of these enzymes can cause a fatty acid oxidation disorder. People with these disorders cannot properly break down fat from either the food they eat or from fat stored in their bodies.

➤ Kansas Newborn Screening for fatty acid disorders

The newborn screening program in the State of Kansas is designed to screen for five different fatty acid disorders. These disorders include:

- Medium chain Acyl-CoA dehydrogenase deficiency (MCADD)
- Very Long chain Acyl-CoA dehydrogenase deficiency (VLCADD)
- Long Chain Hydroxy Acyl-CoA dehydrogenase deficiency (LCHADD)
- Trifunctional Protein Deficiency (TFP)
- Carnitine Uptake Defect (CUD)

➤ Why is newborn screening done for fatty acid disorders?

Newborn screening is done for fatty acid disorders so that babies with these conditions can be diagnosed and treated quickly. Immediate diagnosis and treatment of fatty acid disorders is important for normal development and health. Without prompt diagnosis and treatment, infants with fatty acid disorders will develop varying degrees of developmental delay or mental retardation, medical complications and may even experience death.

➤ Does a positive result from the Kansas Newborn Screening Lab mean that my baby has a fatty acid disorder?

No, not necessarily. Newborn screening identifies babies at increased risk for having one of these disorders. Additional tests will need to be done to determine if the baby actually has a fatty acid disorder.

➤ What are the signs and symptoms of fatty acid disorders?

The age that symptoms start and the types of symptoms that a person has vary. Seemingly well infants and children can suddenly develop low muscle tone, seizures (caused by low blood sugar), liver or heart failure, coma, and death if not treated promptly. The condition may present like sudden infant death syndrome (SIDS).

➤ Is there a cure for fatty acid disorders?

No, there is no cure for fatty acid disorders. However, the outcome is best in infants who are treated early and continue with lifelong treatment. Treatment usually consists of a low fat diet and sometimes medications or supplements. It is most important that children avoid fasting.

➤ Where can I get additional information?

Kansas Newborn Screening at www.kdheks.gov/newborn_screening/info_parents.htm

Save Babies Through Screening Foundation at www.savebabies.org

Screening, Technology and Research in Genetics at www.newbornscreening.info