Organic Acid Disorders
Information for Parents

➢ Overview

Organic acid 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 process protein from the food we eat for use by the body. Problems with one or more of these enzymes can cause an organic acid disorder. People with organic acid disorders cannot break down protein properly. This causes harmful substances to build up in their blood and urine. These substances can affect health, growth and learning.

➢ Kansas Newborn Screening for organic acid disorders

The newborn screening program in the State of Kansas is designed to screen for nine different organic acid disorders. These disorders include:
- Isovaleric Acidemia (IVA)
- Glutaric Aciduria type I (GA-I)
- Multiple carboxylase deficiency (MCD)
- Propionic Acidemia (PA)
- Beta ketothiolase deficiency (BKT)
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-hydroxy-3-methylglutaryl CoA lyase deficiency (HMG)
- Methylmalonic acidemia - mutase deficiency (MUT) or vitamin B12 disorders (Cbl A, Cbl B)

➢ Why is newborn screening done for organic acid disorders?

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

➢ Does a positive result from the Kansas Newborn Screening Lab mean that my baby has an organic 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 an organic acid disorder.

➢ What are the signs and symptoms of organic acid disorders?

The age that symptoms start and the types of symptoms that a person has vary. A newborn affected with an organic acid disorder is usually well at birth and for the first few days of life. If the condition is not treated, babies usually develop vomiting, poor feeding, neurologic symptoms such as seizures and abnormal muscle tone (floppy or stiff), and excessive sleepiness. This can progress to coma and death.

➢ Is there a cure for organic acid disorders?

No, there is no cure for organic acid disorders. However, the outcome is best in infants who are treated early and continue with lifelong treatment. Treatment usually consists of a special diet and sometimes medications or supplements.

➢ 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

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