

MAPLE SYRUP URINE DISEASE [MSUD]

What is MSUD?

MSUD is short for maple syrup urine disease. Untreated children with this condition have a “sweet smelling” (similar to burnt sugar) urine from where its name is derived. In addition, untreated children may have complications such as convulsions, mental retardation and death. It is one of the conditions that can be detected through newborn screening. Early detection can prevent brain damage.

What Causes MSUD?

Due to a lack of an enzyme (BCKAD), some amino acids cannot be broken down and are accumulated in the body producing the signs and symptoms of MSUD.

MSUD is an inherited condition. The BCKAD gene is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the BCKAD gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with MSUD have one working and one non-working gene coding for BCKAD. They do not manifest the disease but can pass them on to their children. They are known as carriers.

If the child inherits the non-working gene from both parents, he or she will have MSUD. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of MSUD?

Children with MSUD may present with lethargy (or sleepiness), vomiting, poor suck, seizures and sweet smelling urine. Without treatment, patients may have swelling of the brain and coma. Brain damage can occur and mental retardation may ensue if patients are not treated well.

How do you diagnose MSUD?

Newborn screening by dried blood spot on a filter paper can detect presence of MSUD in newborns. A blood test called amino acid chromatography is diagnostic of MSUD. A urine test called organic acid chromatography is supportive to amino acid chromatography.

What is the treatment of MSUD?

The main treatment of MSUD is through control of the diet. If the child is well or does not have any illness, he/she should continue taking the special milk formula and be on low protein diet as ordered by pediatrician or metabolic specialist. It is important that the diet be followed to avoid any complications.

What should I do when my baby is unwell?

Children with MSUD may have a metabolic crisis which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drunk well or during stressful events (such as surgery and severe infection). Child may present with lethargy,

seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

