Maple sugar urine disease essay sample

Maple Sugar Urine Disease or MSUD is a genetic amino acid disorder in which body isn’t able to break down the amino acids found in proteins such as leucine, isoleucine and valine. MSUD is also referred to as branched chain ketoaciduria. BCKADs or Branched-Chain Alpha-Keto Acid Dehydrogenase are enzymes that break down those amino acids. This causes the urine to smell like maple syrup. When these amino acids bulld up In the blood they cause complications. These amino acids are found In foods like beans, cheese, turkey, wild game, sausage and milk.

In fact, babies born with MSLJD will drink a manmade formula that is isoleucine-free, leucine-free, and valine-free for the dietary management of maple syrup urine disease. The most common form of Maple Sugar urine Disease is Classic MSUD. All subtypes of MSUD: MSUD Type 1 b, MSUD Type II and Thiamine Responsive MSUD vary in its severity and characteristics of the disease can all be caused by mutations to the BCKD complex. MSUD cannot be contracted; It Is an Inherited disease that you are born with.

A child who has been born with MSUD must have inherited a defective copy of the gene from ach parent, mother and father. The chance of parents who have tested positive for the defective gene responsible for MSUD children have a 25% chance of getting the disease and a 50% chance of being a carrier. The symptoms of MSUD will appear in a newborn after its first encounters with protein (milk). The first symptoms may include a poor appetite, weak suck, weight loss, high-pitched cry or urine that smells “ ke maple syrup or burnt sugar. Babies with MSUD have episodes of Illness called metabolic crisis.

This is a serious health condition caused by low blood sugar and the buildup of toxins in the blood. The first symptoms of metabolic crisis in babies are; extreme sleepiness, sluggishness, irritability and vomiting. The symptoms are similar to an adult with the disease which include; avoiding food, lethargy, vomiting and urine that smells like maple syrup. Diagnosing Maple Syrup urine Disease must be done quickly and accurately for the sake of the patient. Almost all states screen newborns for this disease within 24 hours of birth. MSUD can only be confirmed by analysis of blood or urinalysis.

A rapid conclusion can ensure access to specialized medical services like genetic counseling and dieticians. Newborns suspected of aving this disorder should be evaluated by a metabolic specialist since confirmatory testing and management are complicated and death may occur if the disease is not treated. There are diagnostic lab tests to confirm or disprove having MSUD. Genetic testing Is offered to famllles who believe they may have the defective gene that causes MSUD for future pregnancies. This DNA testing isn’t necessary to diagnose the child.

A prenatal diagnosis involves testing the amniotic fluid surrounding the baby in the womb. They test for enzyme activity in cultured aminocytes or chorion villus cells that are rich in stem cells or mutation analysis. Newborns are screened by a blood sample taken from the heel and analyzed for high leucine levels. A urine amino acid test will reveal signs of ketosis, a high level of ketone bodies and acidosis, metabolic specialist and a dietician. It cannot be stressed enough that babies with this disease need prompt treatment to avoid the serious medical problems associated with it.

The dietician will prescribe a low protein diet or food plan. This restriction must be followed for life. Because foods that are high in branched chain amino acids, like fish and cow’s milk, put children in risk of metabolic crisis. There re medical foods substitutes that may be prescribed by the doctor like baby formula and supplements like thiamine. It is also important to know that children with MSUD must have regular blood tests to measure their amino acid levels. Blood test results may show the need to adjust the diet.

Be advised that newborns with Maple Sugar Urine Disease appear normal at birth. However it Just seems that way for a few days. Plasma leucine begins to rise, usually about 24 hours after birth. The baby may show symptoms of poor feeding. Within a few days ketoacids will appear in the urine, giving it the sweet maple odor to the urine. The symptoms progress from poor feeding to vomiting and lethargy. High pitched cries with spasticity or stiffness are additional signs of high levels of amino acid build up in the blood.

This causes a rapid degeneration or breakdown of brain cell and death if left untreated. The complications associated with MSUD are; coma, neurological damage, seizures, spasticity, blindness in some babies and death. During metabolic crisis treatment involves intravenous fluids, sugars and possibly fats. If levels are too high the patient may be given a solution through IV that helps the body use up excess leucine, isoline nd valine for protein synthesis. Peritoneal dialysis or hemodialysis can be used to reduce the level of amino acids.

Liver transplant surgery is an optional treatment because the BCKD agent that causes the disease resides in the liver. With treatment both prompt and lifelong, children with MSUD can live healthy lives and grow to adulthood with typical development. Early treatment helps to prevent brain damage. It is possible however that even with monitoring and dietary treatment, stressful situations and illness can still cause the levels of certain amino acids to rise. Patients and parents of patients with Maple Syrup Urine Disease should be aware and cautious.