Genetics Newborn Screening Program

Family Fact Sheet

MAPLE SYRUP URINE DISEASE (MSUD)

Protein is an important part of our diet. Protein can enhance body growth and strength when enzymes break the protein down into its proper parts. Babies who have the disorder called maple syrup urine disease (MSUD) cannot break down the amino acids leucine, isoleucine, and valine. Babies with MSUD have high levels of leucine, isoleucine, and valine in their body. With this condition, the baby's urine may smell like maple syrup. If MSUD is not treated, the high levels of leucine, isoleucine, and valine can cause mental retardation, seizures, and other physical problems.
Connecticut began screening for MSUD in 1993. In Connecticut, about 1 in every 250,000 to 400,000 newborn babies has MSUD.

How does a baby get MSUD?

MSUD is an inherited condition. Both the mother and the father have one altered gene that will not allow the body to break down or use leucine, isoleucine, and valine. A baby has to receive this gene from both the mother and the father to have this condition.

How is a baby tested for MSUD?

Prior to discharge from the facility of birth, a small sample of blood is taken from the baby's heel. The sample is then sent to the State laboratory to screen for MSUD. Abnormal screening results are reported to the Department of Public Health, Genetic Newborn Screening Tracking Unit nurse consultant. The DPH nurse consultant will notify the baby's primary care provider of the need for a prompt referral to one of the Genetic Regional Treatment Centers for confirmation testing and follow-up treatment if necessary.

What is the treatment for MSUD?

The Regional Treatment Center specialists will prescribe treatment for MSUD if necessary, which includes a special diet restricted in amino acids leucine, isoleucine, and valine and use of a specialized metabolic formula. Early diagnosis of MSUD leads to early treatment, which is important for the health of your baby.

The information provided is offered for general informational and educational purposes only. It is not offered as and does not constitute medical advice. In no way are any of the materials presented meant to be a substitute for professional medical care or attention by a qualified practitioner, nor should they be construed as such. Contact your physician if there are any concerns or questions.

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