NEWS RELEASE

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NEW STUDY SHOWS METABOLIC DISORDER IN INFANTS MORE COMMON THAN PREVIOUSLY THOUGHT

Nashville, (TENN) - A metabolic disorder that has been linked to abnormal blood sugar levels, heart disease and even sudden death in infants is far more common then previous research had shown, and can be treated with expanded newborn screening reports a study in the September issue of The Journal of Pediatrics.

Dr. Arnold Strauss, James C. Overall Professor of Pediatrics and medical director of Vanderbilt Children's Hospital, is co-author of the study that shows a type of genetic metabolic disorder is five times more common than previously suspected. The study also proves that expanded newborn screening can detect this disorder and potentially keep children from becoming ill.

"We used to think that this disorder, VLCADD, or Very long-chain acyl-CoA dehydrogenase deficiency, was present in on about one in 200,000 births," said Strauss. "But this research shows it's one in 40,000; five times more common."

In the study, eight babies had expanded newborn screening tests that showed an abnormal amount of unusual fat. That elevated fat level is a known indicator of VLCAD-deficiency. This abnormal accumulation of fat happens because the VLCAD deficiency blocks breakdown of such fats that are essential for heart and liver function. Genetic tests defining the mutations (gene abnormalities) in the VLCAD gene were then done.

Six of those babies in this study had the types of mutations previously known to make babies sick, but because the babies were tested and detected early, dietary changes and treatment to prevent illness were made right away. None of those babies has become ill.

"It's so early in the game that we don't know yet what VLCAD disorders typically do to these kids, but the more serious cases are linked with cardiomyopathy, or even SIDS," said Strauss. "What happens is that if the child doesn't eat well, the body is starved or fasted, and they can't use the fats in their blood stream properly. They die from the buildup of these abnormal fatty acids very quickly; they can't go without food for 12 hours."

The chances of death from VLCAD appear to diminish with age, because much like a diabetic, the older children and adults learn to regulate their diet to get enough sugar.

Strauss says the study, titled "MS/MS-based newborn and family screening detects asymptomatic patients with very long-chain acyl-CoA dehydrogenase deficiency" was

possible because of the recent advent of expanded newborn screening. In this research, Tandem Mass Spectrometry (also called TMS or MS/MS) was found to accurately diagnose VLCADD. In TMS, a small sample of blood is used to analyze and detect hundreds of different molecules, including fats and amino acids. The levels of different fats can then be measured and abnormal levels can indicate disorders like VLCADD.

Currently only five states use TMS to test for VLCAD in newborns. Tennessee has recently purchased the equipment to perform TMS and next year will decide which disorders to test for.

Lead author of the study is Ute Spiekerkoetter M.D., formerly of Vanderbilt Children's Hospital, now of Heinrich Heine University Hospital, Duesseldorf, Germany. Co-authors include Strauss and Bin Sun, M.D. of the Vanderbilt Children's Hospital, Thomas Zytkovicz Ph.D. of New England Newborn Screening Program and the University of Massachusetts Medical School, Ronald Wanders Ph.D., of the University of Amsterdam, Emma Children's Hospital, and Udo Wendel M.D. the Heinrich Heine University Hospital, Duesseldorf, Germany.

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