

Babies to be tested for metabolic illness

St. Mark's first in state to screen in standard care

By **Lois M. Collins**

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St. Mark's Hospital will begin today to routinely screen all newborn babies for 30 different metabolic disorders, the first hospital in the state to offer the tests as part of its standard care.

The metabolic conditions are rare but can have devastating — even deadly — consequences if not detected and treated early.

Current state law requires that babies be screened for four metabolic disorders and have a hearing test. The Utah Department of Health is in the process of amending its newborn screening regulations to include the expanded panel, based on recommendations from both its genetics and health advisory committees, according to spokesman Steve McDonald.

Thirty states already require expanded newborn screening, and a March of Dimes report last year criticized Utah for not requiring the tests.

The Supplemental Newborn Screening by tandem mass spectrometry detects more than 30 metabolic disorders, which impact how or if the body breaks down compounds such as proteins, fats and carbohydrates to be used as energy or to promote growth or healing. They include amino acid disorders, organic acid disorders and fatty acid oxidation disorders.

The best-known is MCADD (medium chain acyl-CoA dehydrogenase deficiency). A baby with MCADD cannot burn fat reserves for energy. "The infant runs out of food to burn for energy and can go into a coma and die," said Deb Reiner, spokeswoman for St. Mark's Hospital.

Studies indicate between 5 and 20 percent of sudden infant death syndrome cases are due to MCADD, she said. If parents know an infant has it, they simply wake their baby during the night for a feeding.

One in about 3,000 babies is born with one of the metabolic disorders, said Noriko Kusukawa, assistant vice president of ARUP Laboratories at the University of Utah. They're all treatable if diagnosed early and correctly. Left alone, they can result in mental retardation, damage to the liver, heart or brain, or even death.

Kusukawa knows of one infant with a metabolic disorder who wasn't diagnosed early enough and now needs a liver transplant.

"Effective early treatment has been proven," she said.

"The cost of treating these, when they do get sick, is so high that it is cost-effective to do this even with a 1-in-3,000" chance of finding a problem.

Last summer, University Hospital began providing the expanded screening to newborns, with parental consent, as part of a pilot project between the hospital, ARUP Laboratories and the health department. The goal was not to vet test efficacy, long established, but rather to see if the expanded testing could be

introduced on a large scale in a cost-effective and efficient way, Dr. Nicola Longo, a professor of pediatrics at the U. and director of metabolic services, told the Deseret Morning News at that time.

Although each condition is relatively rare, no one knows which child to screen for which condition until screening answers that question. So testing for all of them makes sense, he said.

From a baby's perspective, it's nothing extra. Babies already have a heel prick to test for phenylketonuria (PKU); the expanded screening is done with blood from the same prick.

Utah currently mandates tests for PKU, galactosemia, congenital low thyroid and hemoglobin diseases such as sickle cell. Health officials are now working with lawmakers and others to expand the requirements.

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