

Benefits outweigh risks associated with newborn screening for disorder

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Newborn screening for a metabolic disorder could lead to false positives -- adding stress to parents, costing money and possibly subjecting a baby to unnecessary follow-up treatment and dietary restrictions.

But the benefits of diagnosing these children early and preventing the risk of mental retardation, disability or death outweigh the costs of a false positive, according to new U-M research published today in the February issue of the journal *Pediatrics*.

"Published studies of expanded newborn screening in a U.S. setting have resulted in favorable cost-effective ratios for screening for this illness but did not include primary data for quality of life effects for a false positive screen," says Lisa Prosser, Ph.D., Research Associate Professor in the Division of General Pediatrics at the University of Michigan Health System and the study's lead author. "Our results show that newborn screening remains cost-effective after accounting for the measured loss in quality of life associated with a false positive screen."

The [metabolic disorder](#) commonly referred to as MCADD - or medium chain acyl-CoA dehydrogenase deficiency -- shows up in one of 15,000 newborns.

When the MCAD enzyme is not working or missing, the body cannot use certain types of fat for energy. This can lead to [low blood sugar](#), or hypoglycemia, and the build-up of harmful substances in the blood. While MCADD patients are usually healthy, repeated episodes of

metabolic crisis can cause permanent brain damage. This may result in learning problems and mental retardation.

Researchers used a [computer model](#) that took into account costs of screening, including the initial screen test, parent time and medical costs of follow-up testing, treatment and the quality of life implications of having dietary restrictions.

They found that screening requires substantial investment of resources but in exchange for important health benefits. The costs of newborn screening for the illness amounted to \$21,000 for every quality-adjusted life year that was gained. A quality-adjusted life year can be thought of as representing a year in perfect health.

"The cost-effectiveness of newborn screening for MCADD is comparable to cost-effectiveness for other childhood interventions, such as vaccinations," Prosser says.

As the number of screened conditions increases, the number of false positives and associated costs also increase, along with the possible identification and treatment of children who may not have experienced related medical problems in the absence of screening, she adds.

"[Newborn screening](#) for MCADD remains cost-effective even when the loss in quality of life associated with false positives or dietary treatments are taken into account," Prosser concludes.

Provided by University of Michigan Health System

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