

YOUR FIRST MOVE? GET HER TESTED

Health Magazine - April 2005

If little Macey Smith had been born a day earlier, she probably wouldn't have survived. The infant has citrullinemia, a disorder affecting 1 in 70,000 children that makes ammonia accumulate in the blood. It is usually impossible to detect until it has caused brain damage or death. But her home state of Louisiana began testing newborns for citrullinemia on November 1, 2004 - the day her blood sample arrived at the lab. She spent 36 hours in intensive care, but the disease was spotted in time. "It's scary to think how close it was," says Macey's mother Laney.

An estimated 1,000 babies would be saved each year if the recent recommendations of a government advisory panel and the March of Dimes are heeded. Both advised that all states screen newborns for 29 treatable genetic diseases. The aim is to standardize a mishmash of state guidelines, says Nancy S. Green, MD, March of Dimes' medical director.

Thirteen states test for all 29 diseases now, while some screen for fewer than 5; others, like Louisiana, are gradually phasing them in. If you're pregnant, check your state's standards at www.genes-r-us.uth.scsa.edu. If your state doesn't test for all disorders, Green recommends having the other tests performed at a private lab. The full battery costs less than \$100.00. Ask your doctor for help finding a lab, or contact Baylor Medical Center's Institute of Metabolic Disease (800-422-9567) and ask about the Supplemental Newborn Screening Project. You'll receive a test kit in the mail. - ES