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Metabolic Birth Defects Screening Begins Early

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Newborns Checked for 29 Disorders

File photo

CHECK UP: The state Department of Public Health laboratory in Frankfort began testing blood samples taken from all newborns in Kentucky's hospital nurseries for 29 metabolic disorders, including cystic fibrosis and sickle cell disease.

FRANKFORT - Statewide screening of newborns for a wide range of metabolic birth defects got under way ahead of schedule last week under a program that could preserve the health and, in some cases, save the lives of an estimated 100 to 120 children a year.

Last Monday, the state Department of Public Health laboratory in Frankfort began testing blood samples taken from all newborns in Kentucky's hospital nurseries for 29 metabolic disorders, some of which can cause mental retardation, organ failure or death. Hospitals must also test newborns for hearing loss.

Legislation sought by Gov. Ernie Fletcher and enacted last spring expanded the state's mandatory testing program from just four disorders to the full 28 recommended by the American College of Medical Genetics and the March of Dimes. Dr. Steve Davis, deputy commissioner of the Department of Public Health, said the state lab will test for one additional disorder because recently installed screening equipment makes that possible.

Screening expanded to six disorders on July 1, Davis said. He said initial plans called for testing to expand to the full range by Dec. 31, but program managers chose to begin it early on a trial basis. On Monday, blood specimens from more than 300 children were screened for all 29 disorders, he said.

Until Dec. 31, Davis said, any blood samples that test positive for any metabolic disorder will be sent to the Mayo Clinic in Rochester, Minn., "to make sure that we're accurate." The state will be "cutting the apron strings loose" at the end of the year, he said.

When the testing and required follow-up with doctors and families are working as envisioned, Davis said, Kentucky could "have what we like to think is the top program in the country" for detecting and promptly treating newborns for genetic flaws that can cause metabolic systems to falter or shut down.

Some Familiar, Some Not

Some disorders that the blood test can detect have familiar names - cystic fibrosis and sickle cell disease among them. Davis said the rest can best be described as disorders of amino acids, fatty acids, organic acids or hemoglobin, plus a few that fit under none of those headings.

By law, a blood sample, usually taken by pricking an infant's heel, must be collected from each newborn between 24 and 48 hours after birth. If technicians at the state lab detect signs of a possible disorder, the child's family will be referred to one of two diagnostic teams, at the University of Kentucky and the University of Louisville, where further tests will be performed.

Davis describes early detection and treatment as crucial. He said nurse case managers working for the program could play a crucial role by tracking down parents quickly when a child's blood sample tests positive for a disorder.

Davis said the testing and follow-up program will initially cost about $4.8 million a year. He said the state would cover $2 million of that amount and would bill hospitals for the rest, at a rate of $53.50 per test.

He said Medicaid will reimburse hospitals for the costs of testing about half the 54,000 children born annually in Kentucky and private insurers will likely pay for others.

Depending on their health insurance plans, families might be billed for part of the cost, but no family will be denied testing or follow-up service because of payment problems, he said.