

In screening babies at birth, state lags

BY NELL SMITH

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Babies born in Arkansas are screened for fewer diseases than newborns in every other state, according to a national group that monitors state newborn screening requirements.

The lack of early checks means Arkansas babies born with certain conditions, such as cystic fibrosis, lose valuable treatment time if they aren't diagnosed until symptoms develop.

Existing state law mandates that all newborns undergo four tests for six conditions, but health officials are exploring the possibility of expanding those screening requirements.

Officials with the state Health Division are considering buying equipment that would allow the public health lab in Little Rock to detect about 20 more conditions. It's an effort that proponents say could save babies' lives and prevent permanent problems.

"If we don't find [babies with these conditions] by newborn screening, some of them will die," said Dr. Stephen Kahler, a pediatrics professor at the University of Arkansas for Medical Sciences who is working with the Health Division on newborn screening. "Some of them will be badly damaged by the first episode of illness. Then we will have found them, and then we will be taking care of somebody who's been damaged."

Presently, the heels of newborns are pricked after delivery so their blood can be placed on absorbent paper. The blood spots are sent to the public health lab to be tested for six conditions: phenylketonuria (more commonly known as PKU), congenital hypothyroidism, classical galactosemia and three types of hemoglobinopathies, including sickle cell anemia. State law also requires cystic fibrosis screening when funding is available, but that has not been done.

Last year, the American College of Medical Genetics recommended 29 tests be done on newborns. The recommendation received the endorsement of the American Academy of Pediatrics and the March of Dimes.

Only the District of Columbia and five states — Iowa, Maryland, Mississippi, New Jersey and Virginia — require all 29 screenings, according to a March of Dimes report released this week. Eight states including Arkansas screen for fewer than 10 conditions. The other states are Kansas, Oklahoma, Texas, New Mexico, West Virginia, Pennsylvania and Montana.

Plans in Arkansas to screen for more conditions are still very preliminary. Health Division employees are trying to determine which screenings to recommend to the state Board of Health, which would make the decision.

The number of tests done in Arkansas will depend in large part on new equipment the state chooses to buy.

A \$ 220, 000 tandem mass spectrometer will give the state the ability to detect a disease known as MCAD deficiency, which causes people to suddenly develop seizures, liver failure or coma. It can also be fatal. The disease affects more than one in 25, 000 babies. With about 37, 000 babies born in Arkansas annually, the screening could be expected to pick up the condition in about 15 babies over a decade.

A tandem mass spectrometer can also detect a number of other far less common diseases as well, including a lethal condition called maple syrup urine disease, so named because of the smell of a patient's urine.

"For MCAD deficiency I think it's pretty clear that the cost-to-benefit ratio is quite favorable,... [the screening] comes out to be a good investment," said Kahler. "And then all the rest of these conditions we screen for are just gravy."

The state's cost for expanding screenings could ultimately exceed \$ 1 million. Officials are considering buying three tandem mass spectrometers for a total cost of \$ 660, 000. Three machines would allow continued operation when one machine breaks down and would accommodate one screening for congenital adrenal hyperplasia that requires a unique machine setting.

Expanding newborn screening would also require funding to educate doctors about what to do if a patient tests positive.

"There's no point in doing any of these tests unless you get information to the right people for the right child in time for them to start the right treatment," said Dr. Dick Nugent, chief of the family health branch of the Health Division. "This is a whole program. This is not just a laboratory test."

Those expenses would be paid by increasing the cost of the test. The current price of \$14.83 for the four tests now done in Arkansas could increase to somewhere between \$50 to \$70 with the additional tests, Nugent said, based on what other states charge.

Hospitals currently pay the screening fee and then are reimbursed by patients and insurers. The state's Medicaid program, which pays for more than half of the deliveries in Arkansas, is required by state law to pay hospitals for the tests. Program officials don't yet know what the financial impact of an expanded testing program will be, said Julie Munsell, spokesman for the state Department of Health and Human Services, which operates Medicaid.

The Health Division has yet to discuss the possibility of expanded screenings with other insurers, said Glen Baker, the public health laboratory's director.

"Medicaid has to... pay for it, but the private insurance companies do not," Nugent said. "So how are we going to deal with that? If the hospitals are going to have to cough up this fee for us to do it, how are they going to get cost recovery?"

That could be a policy question Health Division officials could take to the Legislature when it meets again in January, he said.

While the idea of screening for diseases early has generated wide support in other states, some ethical concerns have surfaced. Some people have expressed concern over false positive results that can unnecessarily worry parents. Some screens pick up abnormalities that may never result in illness and not every condition that could be screened has effective treatments.

Kahler said that those issues have not been overwhelming concerns in other states that have expanded screenings. That's because the screenings are a first step, not a final diagnosis.

"The screening tests are just that," he said. "They say we found something. Let's check into it a little bit further and see what this leads to."

Newborn screening Arkansas screens for the following conditions:

- Phenylketonuria Incidence: Greater than 1 in 25, 000 Disorder resulting from an inability to properly process the essential amino acid phenylalanine. Can cause severe mental retardation if not detected early. Can be treated with a low-phenylalanine diet throughout childhood to prevent retardation.
- Hypothyroidism Incidence: Greater than 1 in 5, 000 Congenital thyroid hormone deficiency that severely retards growth and brain development. Can be treated with oral doses of thyroid hormone to prevent those problems.
- Galactosemia Incidence: Greater than 1 in 50, 000 Disorder resulting from a missing liver enzyme. The condition can cause blindness, severe mental retardation and death. Babies diagnosed with this condition must not have any milk or dairy products in their diets to reduce developmental problems.
- Hemoglobinopathies Incidence for sickle cell anemia: Three types, including Greater than 1 in 5, 000 sickle cell anemia. Sickle cell anemia is a blood disease that can

cause severe pain, vital organ damage, stroke and childhood death. Can be treated with penicillin beginning in infancy to reduce adverse effects. Other tests that state health officials are considering requiring are those that would screen for:

- Cystic fibrosis Incidence: Greater than 1 in 5, 000 Disorder that causes lung and digestive problems and death at an average age of 30 to 35 years old. Treatment, which can improve babies' growth, can include high calorie diet, vitamins, medication and respiratory therapy.
- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency Incidence: Greater than 1 in 25, 000 Disorder that can cause seizures, liver failure, coma and death. Treatment includes nutritional supplements and avoiding long periods between eating.
- Congenital adrenal hyperplasia Incidence: Greater than 1 in 25, 000. A set of disorders resulting from defects in the synthesis of hormones produced by the adrenal gland. Can cause life-threatening salt loss and masculinization of the genitals in females. Treatment includes salt replacement and hormone replacement.

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