

Expanded newborn testing designed to find maladies

By Julie Sevrens Lyons
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When the twin boys were born June 10, they certainly seemed healthy. Each had 10 fingers, 10 toes. Healthy weights. Hearty wails.

Like all other babies born in the state, they had a heel pricked and blood drawn to determine if they have sickle cell disease or a few other types of hidden disorders. But the boys also were included in a trial run of a new state program that will soon test nearly every baby in California for more than 70 serious genetic conditions.

The goal: to get children early

treatment and save lives.

The majority of the disorders are extremely rare. Many parents have never heard of them. Experts predict the expanded testing will turn up only an additional 135 babies a year who have one of the potentially life-threatening conditions.

So when the twins' initial lab results came back indicating abnormal findings, health officials were a bit surprised. And so were the parents.

Within days, additional tests confirmed that the fraternal twins born



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Blood from 1-day-old Anthony Song at Stanford children's hospital will be tested for disorders.



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in San Francisco have a metabolic disorder that occurs in less than one per 100,000 births and can cause severe mental delays if left untreated.

Starting around Aug. 1, the program will be mandatory for all 560,000 babies born in the state, except those whose parents have religious objections.

Critics say the testing has been too long in coming.

California has lagged behind the majority of states in providing the expanded testing. Many parents didn't discover their children had disabling disorders until after symptoms started to appear. By then, it can be too late.

"The tragedy has been that up until now these babies have not been diagnosed," said Dani Montague, director of the California chapter of the March of Dimes. "When they're not treated, babies can suffer from lifelong health consequences such as severe mental retardation, and many of these babies die."

Even with an early diagnosis and treatment, some babies may not survive.

The decision to offer the ex-

panded testing came after years of debate, a commitment by the state to help pick up the tab and the signature of Gov. Arnold Schwarzenegger, making it California law.

One concern had been that the testing, which will cost parents or their health insurers \$78, isn't accurate enough, with some cases going undetected despite the expanded screening. Other parents will be told their child may have a disorder when they do not.

Such false positives "generate unnecessary anxiety on

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the part of parents," conceded Dr. George Cunningham, chief of the genetic disease branch for the California Department of Health Services.

When test results are abnormal, additional blood tests are required to diagnose or rule out a disorder's presence.

During the program's trial run, Cunningham said it has become evident that prema-

ture babies are especially prone to false positives for one of the conditions, congenital adrenal hyperplasia, a genetic defect of the adrenal glands that is treated with hormones.

Public health officials are tinkering with the testing system to reduce the likelihood that small babies will have so many abnormal lab results. They are also working to ensure that results will be available in a timely fashion so that children can receive prompt treatment.

Even with all its flaws, the expanded testing is one of the best tools in medicine to help many of these kids, said Dr. Greg Enns, director of the biochemical genetics program at Lucile Salter Packard Children's Hospital at Stanford.

Enns, who is treating the twin boys from Oakland recently diagnosed with a metabolic disorder, said it could have been weeks or months before they started to show signs that something was wrong. Even then it could have taken some time to determine exactly what was at

the root of their medical problems.

Because of the expanded screening, their condition was caught early enough that they have an excellent prognosis. There are no signs they have experienced any mental retardation, which is one of the hallmarks of their disorder, known as maple syrup urine disease. They are also responding well to their treatment, a low-protein diet and a daily dose of the vitamin thiamine.

Enns expects they will be released from the hospital Friday.

Their mother, who spoke on condition that her name not be used, said she shudders to think what could have happened if they weren't included in the trial run of the new program.

"There's the argument that these things are so rare, is it really worth testing for?" she said Tuesday. As she looks at her 18-day-old infants, she said, "Here's one case where my husband and I would assuredly say, 'Yes, it was worth it.'"

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