

HEALTH

## A baby's first test



Ross Komagome, 11, and his sister Taylor, 2, both suffer from a rare genetic disorder discovered during screening at birth. Now the state is testing all newborns for seven rare disorders, not just two, as in the past. The new law went into effect July 1.

### Newborns now are screened for 7 rare disorders

By Beverly Creamer  
Advertiser Staff Writer

In the excitement of her son Ross' birth 11 years ago, Donna Komagome thought little about the heel-prick blood test her baby received. What's more, she'd never heard of PKU — phenylketonuria — a rare disorder that prevents the body from processing protein correctly and can lead to mental retardation if untreated.

"You don't really ask exactly what they're testing for," says Komagome, remembering the quick blood test given babies before they leave the hospital.

"You don't realize they're testing for some really vital disorders." But a day later the Komagomes learned that Ross was among a handful of Hawaii babies with the rare genetic defect. Then Taylor, 2, was born with PKU. It's changed the Komagomes' lives totally.

"We have to track everything they eat," says Komagome. "If they eat normally, the protein would build up to where it would affect the brain cells."

Hawaii has mandated newborn screening for two rare disorders — PKU and hypothyroidism — for many years. But as of July 1, five more disorders, including sickle cell anemia, were added to the Newborn Screening Program required by state law. (See list on Page C3.)

Screening for PKU began in 1966; congenital hypothyroidism was added in 1983; and then compulsory tracking in 1985 after a little girl with PKU was missed, suffered retardation, and was the subject of a multimillion-dollar lawsuit.

Each of these diseases is "relatively rare, but if your child has it, we really will make a significant difference," says Chris Matsumoto, newborn screening program coordinator in the Health Department.

"It's especially important for our home-birth population, to get the word out to them."

"The additions as of last week make Hawaii one of the most advanced states in the nation in newborn screening."

"Compared to the rest of the country, we weren't doing very well," says Matsumoto. "Now we'll



Suzanya Barton gives her new son Douglas a pig, as a lab assistant picks the baby's feet, and squeezes a few drops of blood onto a card for routine newborn screening. With her first baby three years ago, Barton didn't know what the tests were for, this time she asked.

be in the top 10 percent in the country for the disorders we're screening for. And we're one of the cheapest in the country."

But up 'til now, we've continued to flirt with disaster, says metabolic geneticist Dr. Berkley Powell, former state consultant to the screening program who left two weeks ago to join the University of Nevada School of Medicine in Reno.

"Certainly we were living on the edge," says Powell. The reason: While Hawaii laboratories performing tests informed doctors of positive results immediately, written reports would be mailed to the state screening program with no particular urgency. With 20,000 births a year in the state, that's a lot of paperwork.

As long as doctors acted promptly, that would be no problem. But that one baby was missed in 1982 because the mother never returned to the doctor's office, and the doctor didn't follow up, according to a state spokesperson.

The state tracking system was instituted to prevent that from happening again. And yet, it was still imperfect, according to Powell.

"It was sometimes weeks or months before Newborn Screening knew there was a positive test," says Powell. "The physicians heard right away... (but) there was a delay in getting the results all centralized to one place, and the delay was often several weeks."

"In three or four of these disorders, if you don't diagnose them within 3 to 5 days, that's the danger period. Those are the ones that come in with overwhelming infections and shock."

The new system should end this uncertainty, with an instant computer link to a single, centralized screening laboratory in Oregon that handles newborn screening for five western states.

After a competitive bid process, the Oregon Public Health Laboratory won the Hawaii contract. The cost

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