

State's newborn screening program saved twins' lives

January 9 2012, By Erin Digitale

(Medical Xpress) -- Like a half-million other babies born in California in 2010, Sophia and Charlotte Gonzales each had a blood sample collected after their birth for the state's newborn screening program. But in this instance, unlike many cases, those few precious drops were the difference between life and death: Without the newborn screening program, the identical twin sisters probably would not have survived their first week of life.

“Any genetic disorder that comes to attention by symptoms is already at a disadvantage for treatment,” said Fred Lorey, PhD, who directs the state's [newborn screening](#) program, explaining its rationale. Although genetic diseases do not usually produce symptoms at birth, that's an ideal time to begin treating them. A quick response can often minimize or completely prevent symptoms, warding off consequences such as mental retardation, developmental delay, neurologic deterioration and death.

The routine is relatively straightforward. Blood samples, on filter paper, are overnighted to one of eight labs around the state that conduct the screening tests for 78 different genetic diseases. When Sophia's test results came back a few days after birth, they showed a rare, serious metabolic disorder. Doctors were already aware that the girls seemed lethargic — they were, in fact, slipping toward comas — and the abnormal results raised an alarm that quick follow-up was needed.

Sophia and Charlotte's disease is so rare that, without newborn screening to alert their doctors, their lethargy would likely have been mistaken as a

sign of sepsis, said Gregory Enns, MD, their biochemical geneticist at Packard Children's. "This is the beauty of the newborn screen," Enns said. "The doctor gets a call from the state lab saying, 'Look at this child more carefully.' A flag is raised."

California's newborn [screening program](#) is larger, both in terms of the number of children tested and the number of diseases screened, than any other in the country — and it's among the most sophisticated in the world. Other states currently screen for between 10 and 50 disorders, with most covering at least the 29 diseases recommended for screening by the federal government. Most developed countries have some form of newborn screening, and global population heavyweights China and India are now initiating programs of their own.

California's program continues to expand, most recently adding Severe Combined Immune Deficiency (commonly known as "bubble boy disease") in 2010. Screening costs, currently \$102.75 per child, are covered by mothers' health insurance or MediCal. For the 750 or so children per year with positive test results, the cost savings from timely diagnosis and treatment often rise into the millions of dollars.

Before the test for Sophia and Charlotte's disorder, methylmalonic acidemia, was included in California's newborn screening panel in 2005, Enns sometimes got reports from families devastated by missed diagnoses. "I know of children who died before the newborn screening," he said. "It's a life changer."

Provided by Stanford University Medical Center

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