

BABIES BORN IN CALIFORNIA TESTED FOR GENETIC DISORDERS

July 14 (ABC7) - A major change this week for babies born in California. They'll now be tested for dozens of genetic disorders that are treatable if caught in the first weeks of life. ABC7's Carolyn Johnson reports.

A small prick to the heel, a tiny cry from baby, a few drops of blood that could change the course of a life. Little Sean Michael Bautista entered this world less than 30 hours ago. He's among the first infants in the state to be tested under the newly expanded newborn screening program. This week, California joined 30 other states in the nation. Instead of looking for just four diseases, the new screen can detect more than 35 genetic disorders

Gregory Enns, M.D., director of biochemical genetics, LPCH: "These are conditions that can cause severe disability, brain damage, even sudden infant death. And now these children are being screened for these conditions as a routine, which is the way it should be."

Dr. Gregory Enns heads the biochemical genetics program at Stanford's Lucille Packard Children's Hospital. His specialty is inherited metabolic disorders.

Dr. Gregory Enns: "Sure they're not as common as ear infections or other common pediatric problems such as asthma, but when you put them all together, about one child in a thousand has a metabolism disorder."

Five-year-old Ysabel Jaquez is one of those children - a little girl who almost didn't survive her first week of life. Ysabel left the hospital a healthy baby, but when she was just 3 days old her body started shutting down. Turns out, she lacks a critical enzyme.

Lisa Jaquez, Ysabel's mother: "Her body was basically breaking down itself and trying to feed herself through muscle tissue and she started to die."

Ysabel's condition is considered quite rare. Her mom prefers to say "rarely detected."

"We could not believe that children with metabolic disorders were diagnosed at the expense of their brains or even death."

But now that's all changing with the expanded newborn screening -- a test the Jaquez family lobbied for after Ysabel's diagnosis.

Lisa Jaquez: "It was something that we had worked on, and hoped and dreamed for, because we did not want another parent to go through what we went through."

There are now seven centers in California focusing on the newborn screening results. This one is managed by Stanford and covers 60 hospitals in Northern California. When a child tests positive, the staff here contacts the pediatrician to explain the results, talk about treatment options and make a referral to a specialist.

"We're now able to look in the eyes of moms and dads and say we caught this, and your child has every chance of having a normal life."

For the Bautista family, the test brings peace of mind.

Michael Bautista, Sean Michael's father: "And if there's something they do find, they can do something about it, there's an opportunity to do something about it."

A simple test that will change lives.