

Something's missing in Texas tests for genetic disorders

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By Dave Fehling / 11 News

All newborn babies in Texas are tested to see if they could develop health problems down the road, but even if the results are all good, that doesn't necessarily mean your baby is all right.

Not long after Connor was born, his parents realized something wasn't right.

"That's the whole thing. You, you think about what might have been," said his mother, Lillian Osowski, "He was horribly ill." Connor had stomach problems, he vomited a lot and his muscles didn't seem to develop.

What did his doctor! s say about it? "They were baffled, completely baffled," Osowski said.

Then, when Connor turned one, his pediatrician had an idea that maybe Connor had a rare genetic disorder. Tests confirmed it. Connor is now an active little boy. But he must take medicine eight times a day and eat special food.

It helps, but at five years old, he struggles with speech and needs extra attention in school.

The very sad reality is that a good deal of this might have been prevented.

All it would have taken was a simple blood test. "Absolutely, simple blood test," said Osowski.

Under state law in Texas, all newborns have their blood drawn.

"Its a very important test," said Osowski.

The blood is sent to a lab in Austin where it's tested for genetic disorders that if untreated, can lead to mental retardation or even death.

Recent advances in genetic testing could mean screening for dozens of disorders.

But many states, including Texas, only screen for about a half-dozen disorders.

Some other states, including Mississippi and New York, test babies for up to 10 times as many disorders as Texas does.

Connor had a disorder for which he was not tested.

"His functioning capabilities would be night and day, in my view, if he'd been tested at birth," said geneticist Dr. Hope Northrup, of UT Health Science Center in Houston.

In theory, Connor may never have had any detrimental effects from this had he been treated.

"In theory, yes, that is certainly possible," said Dr. Northrup.

Dr. Northrup oversees little Connor's care.

"So we really are behind a lot of other! states right now," she said. Being behind means not catching many of the disorders before they cause permanent brain damage.



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A simple blood test might have changed Connor's life.

But that is about to change. By this time next year, Texas will begin testing babies for 27 different disorders, including the one Connor has.

Each year, dozens more parents in Houston will be able to learn right away if their newborns have hereditary disorders.

"The test is usually a preliminary result that said that the child may have something, so now a more definitive evaluation is going to be needed," Dr. Northup said.

Counselors will guide worried parents to the right specialists. But there's concern that Texas may have under-funded the program and that critical follow-up care may not be available.

"We are very concerned about that," said Dr. Arthur Beaudet, Baylor College Of Medicine.

Still, the expanded testing is considered a huge step forward that in the long run, will save millions of dollars that now go to care for special needs kids.

Those are the kids whose quality of might have been vastly improved if they had been diagnosed at birth.