LIFE SAVING RULE CHANGE PAYS OFF IN JUST TWO DAYS

Increased blood tests find rare ailment

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By John Pope - Staff Writer

A Opelousas infant named Macey Smith owes her life to an arcane revision of the state's health rules - and a bit of serendipity.

On Nov. 1, Louisiana began requiring its state lab to check for eight potentially deadly genetic disorders, increased from three, in the blood of all newborns. That is in addition to the tests already being done to detect sickle-cell anemia and an underperforming thyroid gland.

The genetic conditions are rare, occurring no more than a few times a year, and they generally involve deficiencies in chemicals the body uses to break down nutrients. Treatment often includes a modified diet.

Macey was born Oct. 24, but her blood sample was eligible for the complete battery because it arrived at the state-run laboratory in New Orleans after Nov. 1.

She was the 57th newborn whose blood was checked for all 10 conditions - and the first to yield a positive result. She tested positive for citrullinemia, one of the new disorders on the list, which could have killed her within weeks if it hadn't been diagnosed. It affects one baby in 70,000 - about one a year in Louisiana where about 65,000 babies are born annually.

The incidence of each of the 10 disorders is low, ranging from one in 100,000 to one in 10,000.

But, "when you combine these conditions that can be detected, you're getting a combined incidence of about one in 5,000. That means we'll come up with about 30 diseases each year," said Dr. Pleasant Hooper, a scientist specialist in the state Office of Public Health's biochemical genetics lab in New Orleans.

Macey's body lacks an enzyme that controls the amount of ammonia in the body. As a result, ammonia builds up in her brain, a condition that would have killed her.

Without tests for such metabolic disorders, parents might make the situation worse by giving infants nutrients their bodies cannot process.

"It does irreparable damage, so it's incumbent on us to identify these babies as soon as we can so they can get the right diet and the right treatment, " said Sandra Adams, executive director of the American Academy of Pediatrics' Louisiana chapter.

Getting information to parents and doctors depends on how quickly each blood sample reaches the state Office of Public Health's lab, Hooper said.

"It takes about 24 hours to know if we have a problem," he said. "If it's a clear concern, we call immediately."

Eventually, the list of metabolic disorders to be checked will swell to 30, but that will be done gradually to keep from overwhelming the lab, which already processes as many as 500 blood tests every day, said Dr. Hans Andersson, a pediatrician who treats Macey at the Hayward Genetics Center at Tulane University Health Sciences Center.

Born at the Right Time

Because the technology lets labs increase the number of conditions for which newborns can be screened, many others are doing so, Hooper said.

"A lot of people have expressed concern that we're not doing all 30 at once," he said. "We discussed this with experts all over the nation, we realized that it was best for us to move in stepwise, to do it safely and do it correctly."

Because of the help Macey has received and will continue to get, "she is, essentially, a normal child," Andersson said.

As he spoke, Macey, swaddled in pink, slept peacefully, and her parents, Laney and Shane Smith, kept looking at her as they spoke of the accidental good timing of Macey's birth.

"It gives me chills when I think how close she was," Laney Smith said.

Like the other metabolic disorders on the list, citrullinemia doesn't have easily detectable symptoms such as a rash or high fever.

But the unchecked buildup of ammonia can bring about poor eating, sluggishness and, eventually, a coma and death, Andersson said.

Before her parent knew about her condition, they said, Macey slept a lot, and they had to wake her up for feedings, but they attributed that to her being born four weeks early.

"I was told premature babies sleep a lot," Laney Smith said.

Lifelong Treatment

They received the news about he test on Nov. 3 and took her to Opelousas General Hospital. The next day, they were referred to Tulane University Hospital, where Macey spent 36 hours in neonatal intensive-care.

Andersson and Amy Cunningham, a metabolic nutritionist at the genetics center, have developed a drug and diet regimen for Macey, who returns to New Orleans for regular check-ups. Medicaid is paying for her care.

Her treatment will be lifelong. "It's a life sentence because it's lifetime care," Adams said, "but it's life, and it can be life with quality."

In Macey's case, both of her parents carried the gene, meaning that each pregnancy had a 25 percent risk of transmitting citrullinemia. However, Macey's 3-year-old brother Brennon is healthy.

Some of these relatively rare genes could be detected through testing, Hooper said, but the process would be difficult, and some conditions might result from mutations, not inheritance.

Rather than investigate causes, Hooper prefers to emphasize treatment, which can be done relatively easily.

Two weeks after Macey's first visit to Tulane, her parents said her condition has improved markedly.

She's more alert, Shane Smith said, and she's ready for a bottle every few hours. She has gained a pound.

"We're delighted," Adams said. The more of these conditions we can identify early, particularly the ones we have good treatment for, the better off we're all going to be."