mandatory miracles

BY HOLLY LEGER

TTUHSC professor's efforts in health outcomes continues to impact newborns worldwide more than 40 years later

Bill and Kerry Scribner of Chalfont, Pennsylvania, walked away from their pediatrician's office on a Thursday last August, proud parents of their second son, James. Just 11 days old, James had passed his weight check in a follow-up to his newborn exam. Despite having jaundice in the hospital, a common condition among newborns, James appeared in good health, and the pediatrician said she would see him again in two months for his routine checkup.

The following day, the Scribners noticed they had missed multiple calls from their pediatrician. Confused, they returned her call, learning the full results from their son's newborn screenings. James' thyroid levels were outside the normal range. She wanted the Scribners to start James on medication right away and take him to the lab the next morning for more blood work.

"It was like a whirlwind," Kerry Scribner said. "We thought it was a little jaundice, and then all of the sudden, I'm like 'What? You want us to start him on medicine? And you're calling the head of endocrinology?' I remember telling my husband, 'This isn't good.'"

Pennsylvania, like Texas, has mandatory testing for newborns requiring a heel stick blood sample in the first 24-48 hours of life. The samples, preserved on special filter paper, are then sent to a lab and tested for genetic disorders. Texas currently screens for 55 conditions, and Pennsylvania screens for 33.

James' second blood test confirmed he had congenital hypothyroidism, a hormone deficiency in which there is partial or complete loss of function of the thyroid gland. Left untreated, it can lead to developmental issues such as mental retardation, learning disabilities and stunted growth. It affects an estimated 1 in 3,000 to 4,000 newborns each year in the U.S.

Knowing nothing about the condition, the Scribners researched it online and started connecting the dots. Infants with hypothyroidism often have jaundice and are sometimes described as "good babies" for how well they sleep.

"Those first two weeks of (James') life, my husband and I were saying to ourselves, 'Oh my gosh! He's a newborn, and he's sleeping like five or six hours at night,'" Kerry recalled. "We just thought that was awesome, not realizing that was all part of the congenital hypothyroidism."

Because it is so difficult to detect congenital hypothyroidism before irreversible damage has been done, all states in the U.S. now require screening for the condition. Pennsylvania began screening for it in the early 2000s. In Texas, efforts for mandatory congenital hypothyroidism screenings were led in 1976 by Surendra K. Varma, MD, DSc (Hon), vice chair of pediatrics at TTUHSC and a pediatric endocrinologist. He also holds the Ted Hartman Chair in Medical Education and is executive associate dean for Graduate Medical Education and Resident Affairs.

Surendra K. Varma, MD, DSc (Hon), a pediatric endocrinologist at TTUHSC, led efforts for mandatory newborn screenings in Texas.

After recognizing symptoms in a Lubbock patient, Varma, worked with then Lubbock Mayor Alan Henry and Texas Sen. Kent Hance, now Texas Tech University System chancellor emeritus, to draft the bill for mandatory newborn screenings. Varma testified in legislative hearings, and in 1977, the bill was passed. Funding was provided during the 1979 legislative session, and by February 1980, screenings began.

Samples were sent to TTUHSC as Varma established the program. After educating others on how to screen and diagnose specimens, he handed the reins to the state a few years later.

Because of Varma's efforts, congenital hypothyroidism can be detected early, and medication quickly administered giving babies like James the chance to live healthy lives.

"Once we diagnose and get the treatment started, I would say 95 percent of them do very good or excellent," Varma said. "As a matter of fact when I follow these patients, and I see them and they are making straight A's and everything is all right, I joke with them that maybe I should start taking the medication."

Beverly Sadler, for one, is thankful for physicians like Varma. Sadler, Bill Scribner's mother, was at a dermatology appointment in Lubbock when she received the phone call about her grandson's diagnosis. Through conversation with her physician, Ashley Sturgeon, MD, assistant professor of dermatology, Sadler learned that Varma was one of her dermatologist's TTUHSC professors.

Sadler contacted Varma to personally thank him for his research and efforts here in Texas.

"I was blown away by that," Sadler said. "They have some of the best doctors in the world in Lubbock. It was very comforting to know they do that kind of research and can get those things passed. I know it took a lot of time and work."

According to the Texas Department of Health and Human Services, the Texas program is now the largest in the world regarding the total number of specimens processed. Between 120 and 150 newborns are identified annually in Texas with congenital hypothyroidism, which means since the program started in 1980, approximately 4,000 to 6,000 babies have received early detection and treatment.

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"It makes me feel great when I look back and see the number of babies this applied to," Hance said. "We saved medical care, we saved medical costs, and we gave each baby a chance to have a full and healthy life."

TTUHSC President Tedd L. Mitchell, MD, said seeing the difference the screening program has made for Texans shows how far the university has come over the last 50 years.

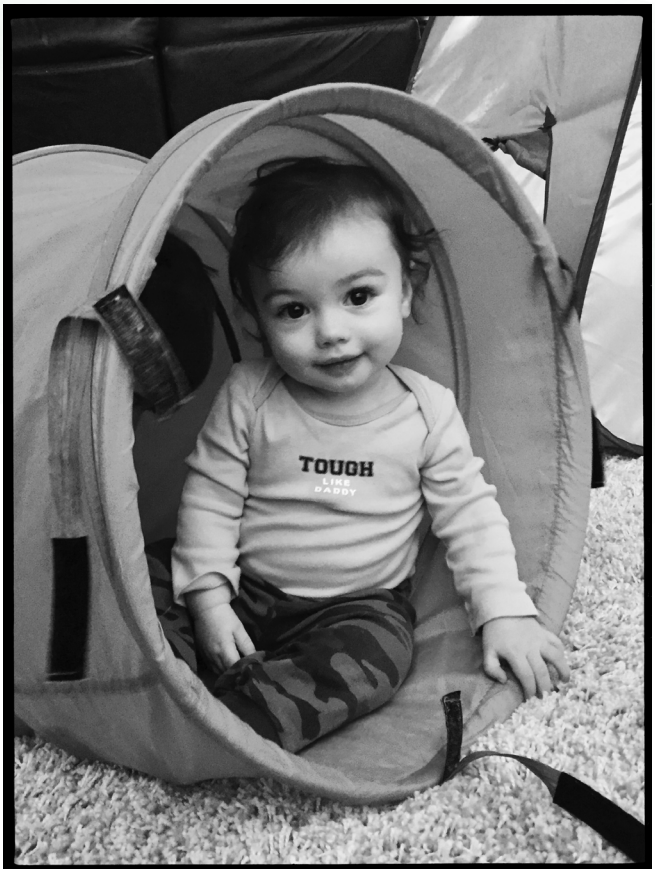
"The impact TTUHSC has on our West Texas communities is profound, and our reach now extends nationwide," Mitchell said. "Not bad for what started as just a little school out in Lubbock."

While much of the developed world requires mandatory screening, other countries lack the technology or government funding for such practices. Varma said he would like to see the benefits of congenital hypothyroidism screening spread to the developing countries as well. In addition to serving children in West Texas and the state, Varma also provides care to children in Vietnam and India as a global health liaison in exchange programs.

"Our problem is the developing countries because access to health care is not good," said Varma, who is a Grover E. Murray Professor and a University Distinguished Professor. "Many of them are delivered at home or in a rural area, and they have not reached screening. So that has been my frustration."

Varma said he was pleasantly surprised to receive Sadler's letter and feels gratified hearing success stories like James'. "Anytime you can make a difference in anybody's life," he said, "it makes you feel very grateful."

Varma responded to Sadler's letter, and Sadler was thrilled to share it with her daughter-in-law in Pennsylvania. Despite living



James Scribner is now a healthy toddler thanks to the newborn screening that detected his congenital hypothyroidism.

The Scribner family: Thomas, Kerry, Bill and James

miles from Texas, Kerry Scribner said she felt incredibly grateful to Varma for understanding the severity of congenital hypothyroidism.

"I feel like he gets it," she said. "It's huge that they're able to diagnosis this at birth and then these children can live normal lives."

Kerry gives her son a thyroid replacement pill every day and will take him to a lab for blood work every six weeks. His treatment will continue throughout his life. She admits the journey has not always been easy. But from month to month, James has continued to hit the milestones expected of babies his age, leaving Bill and Kerry optimistic and grateful.

"Yesterday, I was playing with James on the floor, and I just looked at him. I know I'm his mother, but he's so smart, and he's so beautiful," she said. "I'm just so thankful that we were able to treat his condition. He's going to now get to contribute to the world in a really special way."