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Strauss Studies Mysterious and Deadly Pediatric Metabolic Disorder



Kileen Hall gives Dr. Arnold W. Strauss a hug as they meet for the first time. Strauss published a study on the same rare disorder Hall's two newborns died from. (photo by Dana Johnson)

Lightning couldn't strike twice. That's what Kileen Hall was told right before the tragic death of her newborn son, Tiger. Tiger had a brother, Storm, who also died within hours of his birth three years before. Both babies were full term, apparently healthy, although doctors thought a fairly simple heart defect was to blame for Storm's death. Storm had a full head of hair and Tiger had big, beautiful hands, yet, barely into their second day of life, both boys suddenly died.

It was Tiger's pediatric cardiologist, Dr. Brad Raisher, who had told Hall that Tiger wouldn't fall victim to the same circumstances Storm had. After all, he had checked him thoroughly after birth. Three days after Tiger's death, Hall kept her appointment with Dr. Raisher, who would have been following up with Tiger.

"I was so angry when I went into that appointment," recalled Hall. "I just wanted to know what had happened. But when I finally saw Dr. Raisher, he said he had heard about Tiger's death, and said it had been haunting him that he'd said lightning wouldn't strike twice."

Hall said it was fate when she stood before one of the few pediatric cardiologists in the country who had trained under Dr. Arnold W. Strauss in St. Louis. Raisher said he believed Tiger, and possibly Storm as well, had died from a metabolic disorder.

Hall took home an alphabet soup of initials that stood for a large group of metabolic disorders Raisher had studied under Strauss, called FAOs or Fatty Acid Oxidation disorders. She went online and searched every single one.

Hall honed in on VLCADD, Very Long Chain Acyl-Dehydrogenase Deficiency. The circumstances were just right: sudden, unexplained death and an inability to resuscitate.

Death occurs when the baby's body goes through the normal blood sugar drop after leaving the womb, it is unable to properly burn fats to survive.

Within months of Tiger's death, Hall sent post-mortem blood samples from both babies straight to Strauss's lab where he was able to identify a gene defect. Both boys died from VLCADD.

That was four years ago. At the time, Strauss and his colleagues were discovering that their research had uncovered a cause of Sudden Infant Death Syndrome (SIDS).

Strauss is now the James C. Overall Professor of Pediatrics and medical director of Vanderbilt Children's Hospital. Last month he co-authored a study in *The Journal of Pediatrics* that shows VLCADD deficiency is much more common than previously thought. The study also proves that expanded newborn screening can detect this disorder and potentially keep children from dying.

"We used to think that this disorder, VLCADD, was present in about one in 200,000 births," said Strauss. "But this research shows it's one in 40,000; five times more common."

In his study, expanded newborn screening in other states turned up evidence of the abnormal fatty acids present in VLCADD; at the time of their screening, none of the babies were sick. A quick follow-up test from Strauss's lab proved they did indeed have VLCADD. Dietary changes were made very quickly. None of those babies became ill.

"It's so early in the game that we don't know yet what VLCADD disorders typically do to these kids, but the more serious cases are linked with cardiomyopathy, or even SIDS," said Strauss. "What happens is that if the child doesn't eat well, the body is starved or fasted, and they can't use the fats in their blood stream properly. They die from the buildup of these abnormal fatty acids very quickly; they can't go without food for much longer than about 12 hours."

Some babies can have milder forms of the disease, and apparently grow normally, but the disease can show up later in the form of sudden heart or liver disease, sudden death after fasting, say, for surgery, or severe muscle pain.

Strauss said the recent advent of expanded newborn screening is propelling his research forward and may end up saving lives. Tandem Mass Spectrometry (also called MS/MS) was found to accurately diagnose VLCADD at birth. Now it's just a matter of convincing states to test for it.

Currently only five states use MS/MS to test for VLCADD in newborns. Tennessee has recently purchased the equipment to perform MS/MS and next year will decide which disorders to test for.

Fate has again had a hand in bringing Hall and Arnie Strauss together. Hall says four years ago when she was trying to search out answers about her sons, she wouldn't allow anyone from Strauss's lab to take messages for her. She always insisted on speaking directly with Strauss, long distance from her home in New Mexico.

She laughed and said he was probably bothered by her persistence. But quite to the contrary, Strauss said this is what his life's work is all about. He was happy to have provided answers to the Halls when they lived in New Mexico.

But recently, the Halls moved to Franklin, quite unaware that Strauss was now at Vanderbilt Children's Hospital. The two were put together again after research for this article located the Halls as members of an organization that lobbies for expanded newborn screening. Strauss eagerly invited Hall to visit his lab at Vanderbilt.

The best part of all this, Strauss said, is that the persistence of parents, like Hall is driving expanded newborn screening forward. If states do opt to use the screening to detect VLCADD and other related disorders, his research may have a chance to make a difference, and to save lives.