TUBA CITY, Ariz. -- Lorria Trujillo never felt she knew enough to question doctors about her 6-month-old daughter's health. She didn't second guess them when they insisted Charlotte merely had a viral infection after months of being sick, nor question them when the girl's lungs collapsed.

Trujillo watched as her baby was unhooked from life support and held her until she died.

That was 1995.

Written on the child's death certificate is "severe infection," but Trujillo now knows the condition that claimed her daughter's life is the same one her 9-year-old daughter, Grace Marie Yazzie, suffers from.

"With most families, it's the mother that's really responsible for taking care of their babies," the Navajo woman said. "I really felt like I didn't do my part as a mother, and I kept looking for something I would have missed. Would I have known?"

Without treatment, children have no chance of surviving severe combined immune deficiency -- a disorder that's more commonly referred to as "bubble boy disease" after a Houston boy who was forced to spend his 12-year life in a plastic bubble free of germs.

In the Navajo population, one in every 2,500 children inherit SCID, a condition that endows them virtually no immune system. In the general population, SCID is much more rare, affecting one in 100,000 children.

Before Grace was born, Trujillo had researched SCID, and knowing that she and her now ex-husband had a one in four chance of having another baby with the condition, she insisted on a blood test. Although doctors didn't think the test was necessary, Trujillo knew not to keep quiet this time around.

"The longer it took, the more apprehensive I got," Trujillo said. "When I saw the doctor come into the delivery room, I could tell there was something wrong. He told me, 'Yeah, she tested positive for SCID.'"

Dr. Mortan Cowan, who has worked with SCID patients for more than two decades, encountered his first case in the mid-1980s when he was asked to watch over Navajo patients for a doctor in Denver
who went on sabbatical.

"I didn't think much about it and said, 'Sure,'" Cowan said. "And we had four kids that year -- all Navajo kids -- who had this disease."

To Cowan, the disease appeared to be linked to genes, so in 1986 he and a research geneticist decided over lunch to find the gene -- a quest that would take 15 years, Cowan said.

"When we ultimately found the gene and went back, we were able to show that it was the same gene mutation in every Navajo and Apache child that had the disease," Cowan said.

American Indians typically have had a higher infant mortality rate than other ethnic groups because of poverty and limited access to medical care, said Dr. Diana Hu, chief pediatrician on the 27,000 square-mile Navajo Nation. So when an infant died of infection, "you don't really notice that is odd," because others also were dying of infection, she said.

Things changed in the late 1970s and early 1980s with improvements to health care.

"This is not just kids dying, this is something odd," Hu recalls thinking. "When you start to lower your infant morality (rate), you start to notice when kids die."

But detecting the disorder wasn't easy. What can frustrate parents are that the symptoms of SCID aren't much different from the common cold or flu. Normal kids can have numerous ear infections in a year but are treated with antibiotics and the infection goes away.

In SCID patients, the infection lingers and worsens.

"I think that there's a learning curve for families, there's a learning curve for us as doctors, there's a learning curve for the Navajo Nation with illnesses like that," Hu said.

Researchers have identified about a dozen genes that cause SCID. Cowan, director of the Pediatric Bone Marrow Transplant Program at the University of California-San Francisco, argues that Navajos and Apaches suffer from the most severe form of the disorder in which they lack a gene called Artemis. Without it, the children's bodies aren't able to repair DNA or develop disease-fighting T cells and B cells.

"These kids are the most difficult to treat," he said.

The autosomal recessive gene found in the Navajo and Apache populations can be passed from one generation to the next without harm. But when two people who carry the gene have children together, there's a one in four chance their children will be born with SCID. The type seen in the Navajos and Apaches is known as SCIDA, because the two groups share a common language root, Athabascan.
The disorder is something Lynnae Redhouse and Sean Frank, a young Navajo couple, never had heard of.

Day after day, their son, also Sean Frank, would cough until he turned blue and sleep more than a baby should. His hands and feet shook, and soon after he was fed, the milk would come right back up.

Redhouse and Frank knew it was normal for a child to occasionally get sick, but something here wasn't right.

"It turned into just a routine of waking up all the time for him because we were so worried he wouldn't wake us up at all," said his father.

Each time Redhouse and Frank would seek care for their son, the message was the same: Take him home, he's fine, doctors told them.

It turns out he wasn't fine, and not until the child was taken to the University of New Mexico Hospital in Albuquerque did his parents find out their son has SCIDA.

"We never thought anything like this would happen to us. In my heart, when I heard that result, we thought, 'No, he's a healthy baby,'" Redhouse said. "He just didn't look like he had SCID. People would say to us, he's not losing weight, he doesn't have skin rashes."

Bone marrow transplants can be a lifesaver for children who suffer from SCIDA, providing them with stem cells that take root and begin producing T cells. But even with the best care, not all children will be saved.

Shortly after receiving a transplant this summer, baby Sean's cells are growing, "just a little bit," Redhouse says, but it could be months before he can return to his home in Farmington, N.M., on the edge of the Navajo reservation.

"He has to have a certain number before he gets to go," said Redhouse from San Francisco, where baby Sean is being treated.

At clinic each week, doctors weigh baby Sean, take his temperature, blood samples, and check for any rashes, changes in behavior or sleeping patterns.

Because SCIDA patients lack the Artemis gene, Cowan and his team have decided not to prep the patients for bone marrow transplants using the standard approaches, such as radiation or chemotherapy, which break down DNA in order to rebuild the immune system.
"It turned out that was extremely dangerous for the Navajo SCID babies," said Jennifer Puck, who studies inherited immune deficiency disorders at UCSF. "In fact, many of them died before the transplant could be given to them because of the toxicity from the radiation treatment."

On a recent day at the Tuba City Regional Medical Center, 8-year-old Justin Knight is playing with action figures as he awaits a bimonthly reunion with two other SCIDA patients.

Inside the infusion room, Trujillo's 9-year-old daughter, Grace, rants about her favorite sports, how much she likes math and what she has learned from having SCIDA nearly a decade after being diagnosed.

"Take medicine and eat the right food," she jokingly says, holding up a candy bar. "Chocolate keeps me going."

A chair opens up nearby, and Justin enters the room.

He and fellow boarding school classmate Joron Mike stare up at the TV, seemingly oblivious to everything else.

Grace and the boys each have a port-a-cath, a direct conduit to a major blood vessel -- implanted in their chest. Prolonging their lives is a two-hour infusion of gamma globulin to reinforce their B cells. Once diagnosed with SCIDA, most patients at the Tuba City hospital are sent hundreds of miles from their homes on the reservation to UCSF Children's Hospital to undergo transplants.

"These kids just didn't happen to fully take with the bone marrow," said Mary Schillo, an infusion nurse at the hospital. "They will have to do this for the rest of their lives."

Grace pulls out a rubber band wrapped around her dark brown pony tail and reveals a white patch of hair. She boasts about a Mickey Mouse-shaped discoloration on her back. Parts of her fingers and around her mouth also are lighter than the rest of her skin.

She asks her mother "how come I'm like this and nobody else is," why she's shorter than other children and says other kids make fun of her, Trujillo said.

"It's kind of hard for her to understand why kids would be mean," Trujillo said. "I just have to tell her, 'It's not your fault. Some kids are different. They think differently.'"

Researchers aren't sure why some SCIDA patients never lose their baby teeth, are shorter than other children or have severe oral and genital ulcerations. One theory is that the lack of Artemis is responsible.

As dinner trays are set down on tables for Grace, Joron, Justin and their family members who
accompany them to the hospital, they are told to wash their hands.

Germs are their enemies.

While most kids are making mud pies and snow men, these children are urged to stay away from things that can trigger an infection. A fever or diarrhea could mean a trip to the emergency room.

Some 3,500 babies are delivered each year at hospitals on the Navajo Nation.

A hospital policy manual that Hu developed outlines what diseases commonly are seen on the Navajo Nation and what to do if a health provider suspects SCIDA.

"Most of us have been here for 10 years and have seen it happen," Hu said. Others, she said, "will have read about this stuff but never seen it."

Cowan said there are continued efforts to educate new doctors on the reservation about what to look for, especially because SCID is diagnosed at a much higher rate on the Navajo Nation than elsewhere.

"Even with that," he says, children "sometimes slip through."

Hu wonders whether children she has seen die would have lived if diagnosed earlier.

"Our kids who are diagnosed earlier and transplanted earlier tend to do better," she said. "Our goal is to spare families from this tragedy."

Although Navajo and Apache children suffer from SCID at high rate, there's no standard test to detect it.

Most children will be diagnosed after a persisting infection, usually within three months of birth, Cowan said.

Mothers whose families have a history of SCIDA can request a prenatal test, but Cowan notes it is expensive and "any time you do some kind of procedure involving a fetus, there's always a risk."

"You couldn't do that as a prenatal screening," he said.

All newborn's heels are pricked, and the blood is tested for a range of genetic diseases that doesn't include SCID.

Puck is trying to solve the problem that not all Navajo children are tested. A special test she is developing would determine whether a child is immune deficient.
"In the nursery they look fine, so there's no way anybody could tell," said Puck, taking a break from writing a grant proposal for the test. "But there's this problem percolating, and it's going to rear its ugly head. So the idea is to do screening early enough so that you can make the diagnosis early enough to have the optimal treatment."

Grace currently lives with her grandmother, Nora Trujillo in Tonalea on the western side of the Navajo reservation. Meanwhile, Lorria Trujillo, a health educator, is in Flagstaff pursuing a nursing degree at Northern Arizona University.

"I've always been interested in the health field but even more so when I had my experience with Charlotte and Grace," Trujillo said. "I just kind of realized that a lot of families got limited information if they had a child with SCID."

Each summer Trujillo travels to Tuba City to attend an annual reunion for SCIDA patients and their families.

For Trujillo, seeing SCIDA patients in their teens and early 20s provides hope that Grace can grow to become a woman. Her 17-year-old cousin, Christopher David, is among the SCIDA patients.

"I guess the main way ... I try to cope with it really is keeping myself well-informed and always making a point to be there when they have that get-together with Dr. Cowan and the other families," Trujillo said. "I need to see the other kids there and that they're doing all right."