Health Matters

Sickle Cell: One Gene, Many Faces, And A Quest For A Cure

(NAPSA)—In 2000, a mysterious disease was making 3-year-old Mercy Mendoza so ill that her grandmother bought a burial plot for her in the little town in Honduras where they lived. Swelling, pain and immobility were fast eroding her health. Finally, a doctor who had been trained in the United States recognized her condition: It was sickle cell disease, an inherited blood disorder.

Thirteen years earlier in India, then 4-month-old Kirti Dasu was diagnosed with the same condition, as eventually was his brother. But while Kirti, now 30, survived, his brother did not. "[The doctors] didn't know much, so they treated Anup [his brother] the way they treated me, and it was fatal," said Dasu, whose parents eventually brought him to the United States for treatment and care.

Around the same time in Des Moines, Iowa, Cassandra Trimnell became the first child diagnosed with sickle cell disease through that state's newly implemented newborn screening program. Her mom was stunned. "Like most others in the African-American community, she didn't know she had the sickle cell trait until her beautiful baby was born," said Trimnell, 31, but she worked hard and got the care Cassandra needed.

The experiences of Mendoza, Dasu and Trimnell—on continents thousands of miles apart—turned out to be life changing: Each is now a fierce advocate for sickle cell disease awareness and education. Their stories speak to the complex journey of the sickle cell trait. They also underscore a little-known truth—that sickle cell disease, the most common inherited blood disorder, has a far-reaching impact on a surprisingly diverse swath of the global population.

The rare condition affects at least 100,000 people in the United States—mainly African Americans, but also Hispanics and Asians—and 20 million worldwide. They share not only a diagnosis but, according to new research,

Although about one in every 365 African-American babies is born with sickle cell disease...

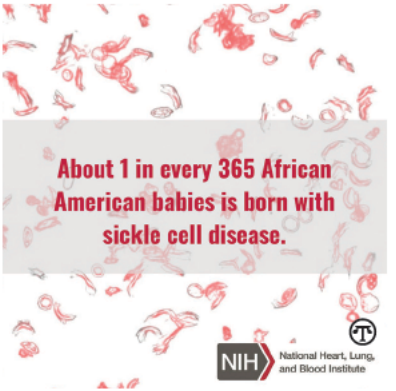
perhaps an ancestor who lived some 7,300 years ago in what is now the Saharan Desert.

The child was born with a genetic mutation that has endured over 250 generations because it protects against malaria, a major killer, then as now. It also causes deformed, sickle-shaped red blood cells. A child develops sickle-cell disease if he or she inherits two faulty copies of the gene, one from each parent.

As Mendoza, Dasu, Trimnell and others with the condition know, the results can be serious. The sickled cells can clog the blood vessels and deprive cells of oxygen. In turn, this lack of oxygen wreaks havoc on the body, damaging organs, causing severe pain, and potentially leading to premature death.

"Fifteen minutes from now, I could have a crisis with chest pain or be unable to breathe," said Mendoza. "There are times I have to go to the hospital, and I don't think I'm going to make it."

The National Institutes of Health (NIH) spends some $100 million annually on sickle cell disease research, which has led to pain-reducing treatments such as hydroxyurea and also laid the groundwork for genetic approaches to potential cures, such as the genetic editing of bone marrow cells. To speed up this genetic research, NIH's National Heart, Lung, and Blood Institute (NHLBI) in September launched the Cure Sickle Cell Initiative, an effort designed to move the most promising of the genetic therapies into clinical trials within five to 10 years.



...there can be a cure. The time has come.

"Through this initiative, researchers will be taking advantage of the growing number of tools they now have to correct or compensate for the defective gene that causes this painful disease," said Keith Hoots, M.D., director of NHLBI Division of Blood Diseases and Resources. The goal, he added, is to find more than one cure because patients respond differently, depending on their ages and other factors. For example, bone marrow transplants—the only cure for sickle cell disease to date—tend to be more successful in children, but only a few patients can find suitable donors. "There may never be just one universal cure," he said.

The initiative spells hope for Mendoza, Trimnell and millions of people living with the condition. And it gives Dasu another reason to continue advocating for new discoveries that help people like him. In 2017, Dasu received a bone marrow transplant and he has made an astonishing recovery.

"I'm good!" said Dasu. "Now, for the first time, I mean it when I say 'good.' Not just 'not too much pain,' but 'good.'" His hope, he said, is that new therapies will make it possible for others to say the same.