Cystic fibrosis diagnosis misses more people of color. This couple seek to remedy that.

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USA TODAY

Published 5:00 a.m. ET Feb 28, 2022

Terry Wright was 54 when he finally understood why he'd been in pain most of his life.

His diagnosis with cystic fibrosis, an inherited lung disease, explained the daily vomiting and stomach pain, regular lung infections and near-constant sinus problems.

Over the past nine years, new treatments have transformed cystic fibrosis, adding years to a patient's life expectancy and quality to those years.

But only if it's diagnosed and treated early in life.

Wright's condition was missed for decades, largely because he's Black.

Only one doctor had ever mentioned the idea that Wright might have cystic fibrosis – which Wright had never heard of – but quickly dismissed the idea because, he said, Black people didn't get it.

Even today, children of color are often diagnosed with cystic fibrosis at older ages than white children, receive specialized care later, are less likely to participate in clinical trials and are less likely to benefit from current medications for the underlying cause of the disease.

Wright and his wife, Michele Wise Wright, have launched an effort to change all that, called the National Organization of African Americans with Cystic Fibrosis. The group has just issued a free, online screening tool, called The Wright Cystic Fibrosis Screening Tool, to call attention to the problem and allow patients to see for themselves if their or their child's symptoms might add up to cystic fibrosis.

Michele and Terry Wright have formed a national foundation to draw attention to the needs of non-white cystic fibrosis patients. *Yancy Hooks*

Terry Wright's story also speaks to a larger problem, experts say. Families of color and those with low incomes are often left behind by scientific advances, particularly when it comes to rare diseases where Blacks, Hispanics, Asians and Indigenous people account for a fraction of an already tiny pool of patients.

Cystic fibrosis is considered a rare disease. There are about 30,000 patients in the United States. Roughly 80% of cystic fibrosis patients are white.

By contrast, sickle cell disease, which has three times as many patients, largely affects people of color. A 2020 study found that cystic fibrosis receives 3.5 times more in federal research funding and 75 times more money from foundations, despite its smaller population.

Black and other minority children are also often left out of clinical trials, the research that leads to treatment advances, which puts them further behind their peers, said Linda Goler Blount, president and CEO of the Black Women's Health Imperative, a national nonprofit advocating on behalf of the health and wellness of Black women and girls.

"The No. 1 reason (they don't participate) is nobody asks them," she said. "Doctors make assumptions about what a patient will or will not do."

Many people of color or low-income backgrounds have overcome incredible odds to find effective treatments for rare diseases, but that's the exception rather than the rule, said Pamela Gavin, executive vice president for the nonprofit National Organization for Rare Disorders, which is working to improve diversity in trials and treatment.

"Those folks are wonderful, beautiful stories of inspiration, but it's certainly not the norm," Gavin said.

Her organization is working with 31 institutions to provide diagnoses and care for rare conditions, and they have made explicit their goal to diversify their policies and represent more diverse populations, including people with different points of view and disability levels, as well as people of color.

"It's important, silent work that needs to happen behind the scenes in order to strengthen the community at large," she said. "We have to be intentional. No longer 'build it and they will come' will suffice. Because it doesn't happen."

Cystic fibrosis advances

Black people have always had cystic fibrosis. The disease was first described by New York pathologist Dorothy Andersen in 1938. Her original case studies included Black and Hispanic children, said Dr. Jennifer Taylor-Cousar, a pediatric and adult pulmonologist who runs an adult cystic fibrosis center at National Jewish Health in Denver.

But those children were ignored, and cystic fibrosis became known as a white disease.

Most of the children in Andersen's case studies didn't survive kindergarten, Taylor-Cousar said. Even by the time that Wright was born in the 1960s, treatments would only have been available to help treat some of his symptoms.

But now, the stakes are much higher.

Treatments have been approved to start as early as 4 months old and are being tested in even younger children, said Dr. Clement Ren, who directs the Cystic Fibrosis Center at Children's Hospital of Philadelphia. "Our vision for the very near future is to be able to diagnose infants in the first two to four weeks of life and start these medications to prevent the complications of CF."

Over the past 30 years, the lifespan for those with cystic fibrosis has been slowly increasing, largely through more aggressive treatment and – more recently – new medications. From 2016 to 2020, the median predicted survival in CF was 50 years, meaning that half of infants born during this time would be expected to celebrate their half-century birthday, even without further progress in treatment.

But those gains aren't shared by Black, Asian and Hispanic patients, whose disadvantages start early and stick, research shows.

Newborn screening, widely available since about 2010, has been a huge advance overall but also may increase disparities of care, said Dr. Susanna McColley, a cystic fibrosis expert at Northwestern University's Feinberg School of Medicine.

Cystic fibrosis expert Dr. Susanna McColley *Janice Terry At Ann & Robert H. Lurie Children's Hospital Of Chicago*

If a child doesn't test positive in those first few days, it may be years before a doctor thinks to run a definitive sweat test to look for cystic fibrosis, she said.

"More kids who don't have northern European ancestry are going to have false negative tests," she said. There's a lot of variability in when children are first seen at a cystic fibrosis center, she said, with Black, Hispanic, Asian and Indigenous people "seen at a later age."

Even when they test positive, Black and Hispanic infants "might present at a lower level of alert," delaying referrals to specialists, Ren said.

Newer medications are transforming care for cystic fibrosis, though the benefits are not equally distributed.

Medications have reduced the number of lung flare-ups from 40% annually among adults in 2019 to 17% in 2020, and lung, liver and kidney transplant rates are all falling among those with cystic fibrosis, according to data in the Cystic Fibrosis Foundation's Patient Registry.

The drug company Vertex Pharmaceuticals has developed treatments that can now help up to 90% of patients with CF mutations. But the 10% still left behind includes a high proportion of people of color because they are less likely to have the common mutations against which most drugs are targeted.

"There is less treatment available for people who are racial and ethnic minorities," McColley said.

The Vertex drug Kalydeco, approved for use in early 2012, has shown a 78% reduction in mortality and 89% reduction in the need for transplants and 50% reduction in hospitalizations over eight years, said Dr. David Altshuler, the company's chief scientific officer.

Cystic fibrosis patients normally lose about 2% of their lung function a year. Based on long-term data, most of the company's medicines cut that rate in half, Altshuler said, but its newest approved drug, Trikafta, showed no decline over two years.

A child with cystic fibrosis gets an inhaler treatment. *Getty Images*

"People with CF are able to go to school, get married. There's been a birth explosion among people with CF," said Fredrick Van Goor, the company's head of Cystic Fibrosis Research.

Vertex's approach to identifying and treating people with rare disease mutations has become a standard across other diseases, Altshuler said. "It's very gratifying working on CF," he added.

Although the drugs are directed at common cystic fibrosis mutations, they also benefit some people with rarer mutations, who are more likely to be people of color, Van Goor said. Terry Wright, for instance, has a rare mutation but said he has seen benefits from taking Trikafta.

A cure may be possible someday too.

In November, the Cystic Fibrosis Foundation announced it was partnering with Flagship Pioneering, to spur the development of genetic-based therapies for cystic fibrosis. The foundation agreed to invest up to $110 million, and Flagship agreed to set up a separate company specifically focused on potential treatments for cystic fibrosis. The partnership is part of the foundation's $500 million Path to a Cure Initiative, to accelerate treatments and drug development for the underlying cause of the disease and ultimately deliver a cure.

Using genetic therapy, researchers may eventually be able to repair or replace the faulty gene that causes so much suffering.

Vertex is focusing its gene therapy effort on the 7% of CF patients who don't make any of the cystic fibrosis protein, and who therefore aren't getting the benefit from existing medicines. Moderna, made famous by its COVID-19 vaccine, is collaborating with Vertex on a gene therapy that researchers hope will induce patients' bodies to make the protein. Human trials are expected to start soon.

Trying to catch up

As a child, Wright was diagnosed with nearly everything except cystic fibrosis. He tried to play sports but was left on the sidelines gasping for air – and was diagnosed with asthma.

His constant stomach pain, vomiting and belly swollen with gas were determined to be ulcers.

He had his gallbladder removed, several sinus operations and pancreatic surgery that lasted eight hours and left him hospitalized for a month. But still, doctors didn't put the pieces together.

"This is what's so frustrating," his wife, Michele, said in a video call. "The signs and symptoms he had were classic."

If doctors hadn't been distracted by his skin color and what they thought they knew about cystic fibrosis, they would have spotted it decades ago, she said. "I believe it could have changed his whole life."

Terry Wright, who wasn't diagnosed with cystic fibrosis until he was 54 years old. *Michele Wright*

He has worked mostly odd jobs because his illness made a career impossible, Wright said. It also made him infertile.

With their national organization, the Wrights hope to bring attention to the problem of missed diagnoses for Black Americans and other minorities, to expand early screening and ensure that minorities are included in clinical trials.

On their screening tool, people answer questions about whether they have symptoms such as frequent, productive coughs, nasal polyps, loose stools, shortness of breath out of proportion with exercise, salt crystals on skin after exercise or male infertility.

Anyone who checks off a number of symptoms should ask their doctor for a sweat test, which definitively diagnoses cystic fibrosis, said Taylor-Cousar, who helped develop the screen. "This tool is meant to help empower patients to make their own diagnosis."

Michele Wright, an engineer with a Ph.D. in public policy, also wrote and directed a short film, "54 Years Late," telling Terry's story and named for his age at diagnosis.

And the duo co-founded the annual Blacks, Indigenous, and Other Minority Ethnicities with Rare and Genetic Disease (BIOMERGD) Conference to increase awareness of rare diseases among people of color.

Terry, 59, is now on medication and aggressive therapy for cystic fibrosis.

He wears a vest 1½ hours a day that helps loosen the mucus in his lungs. Enzymes help him digest his food.

Although Trikafta was first approved for people with more common mutations, its use was expanded to allow people like Wright to try it. On it for about a year, he thinks it's making a difference.

"I'm not wheezing in my lungs like I was," he said. "I believe I am getting some help."

But Wright still carries the emotional scars from a lifetime of illness.

"It would have been wonderful if I had had a name for what I was dealing with," he said. "There were things I could have done myself. I think a person does need to take their own health in their hands."

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Health and patient safety coverage at USA TODAY is made possible in part by a grant from the Masimo Foundation for Ethics, Innovation and Competition in Healthcare. The Masimo Foundation does not provide editorial input.