New drugs for cystic fibrosis bring hope

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Emily Schaller likes to travel as part of the cystic-fibrosis advocacy she does, but the daily regimen she uses to manage her disease weighs her down.

"You have to bring three extra bags with meds and the vest," she said.

Schaller, 28, takes five or six pills every time she eats -- as many as 40 pills a day -- to give her body the digestive enzymes it needs to absorb nutrients. She also uses an inflatable vest that attaches to a 20-pound compressor and does separate breathing treatments twice a day to help clear mucus from her airway.

Still, Schaller considers herself lucky. She's part of the growing group of people living longer with cystic fibrosis, a disease that until recently meant certain death before high school.

Diagnosed when she was a year and half old, Schaller is one of 30,000 Americans who have the inherited disease, which typically affects the lungs and pancreas.

There is no cure for cystic fibrosis and there have been no big breakthroughs, but incremental treatment advances are paying off in increased life expectancy and greater quality of life for many patients, experts said.

"The children born now (with cystic fibrosis) will probably live into their 40s or 50s," said Dr. Jerry Nick, director of the adult cystic fibrosis

program at National Jewish Health in Denver. "A combination of a lot of small improvements has made a huge difference."

In 2007, nearly half of patients survived past age 18, up from only 28 percent who lived that long in 1985, according to the Cystic Fibrosis Foundation.

New drugs now in development also are spreading hope that one day they'll be able to correct the root cause of cystic fibrosis -- flaws in the CF genes inherited from each parent. Four drugs targeting the fundamental cause of CF made by Vertex Pharmaceuticals, Pharmaxis, PTC Therapeutics and Inspire Pharmaceuticals are in phase 3 clinical trials, said Laurie Fink, spokeswoman for the Cystic Fibrosis Foundation. In total, there are 30 drugs in the pipeline targeting various aspects of the disease.

The Cystic Fibrosis Foundation has what Fink described as a venture-philanthropy business model that's unique to disease-mission organizations. Founded in 1955, it straddles two ends of the patient experience. Because cystic fibrosis is considered a rare disease, the foundation hedges some of the risk for pharmaceutical companies by funding early-stage drug research. It also accredits 110 cystic fibrosis care centers across the country, oversees their quality and tracks patients' progress in a voluntary registry.

Cystic fibrosis causes thick, sticky mucus to build up and clog patients' respiratory tracts, causing what can be life-threatening infections. Persistent coughing is a common symptom.

"You turn red and people think you're dying, but we're used to it," Schaller said of CF patients. "When someone gets pneumonia or bronchitis, that's kind of how we sound pretty much all the time."

But a host of antibiotics and inhaled treatments are preventing more patients from suffering some of the disease's most serious complications, and there is a wide range of health among patients, said Dr. Susanna McColley, director of the Cystic Fibrosis Center and head of the division of pulmonary medicine at Children's Memorial Hospital in Chicago.

"We have a large number of therapies we prescribe to people who are in good health to try to prevent pulmonary exacerbations," she said.

Improvement in treating patients' nutritional problems counts as one of the biggest advances, she said. "Having everyone adopt this high-calorie diet has in and of itself saved hundreds of years of life."

Vitamin supplements also play a role in shoring up faulty nutrient absorption, Nick said. "Back in the old days, children would die in infancy of malnutrition."

Patients are expected to live to age 37 or 38 on average now, up from age 29 in 1992, said Dr. Mary Ellen Kleinhenz, professor of medicine and director of the adult cystic fibrosis program at the University of California at San Francisco. Healthy patients typically come to UCSF four times a year for a check-up.

Compared with the medical protocol 20 years ago, "we're better organized in how we deliver care to patients with cystic fibrosis," Kleinhenz said. "People are coming of age with more normal lung function, more normal weight and height, so they're healthier."

Still, self-care is often time-consuming and expensive, and not all patients want to do everything possible to take care of their health, McColley said. Doctors can provide clear explanations and time-management advice for treatments, but it's up to patients to decide how

much they can handle, she said. "When you take your inhaled therapy and do chest therapy, it's like money in the bank because you're saving health for later."

Finances are another burden. Although programs exist to help uninsured and underinsured patients afford their medications, some require a lot of paperwork or aren't available quickly when needed, she said. "I have families who have copays of $1,000 a month for medication."

Since Schaller joined a clinical trial last December and started taking a new pill made by Vertex, she said her lung function has risen to between 80 percent and 90 percent from 70 percent.

"It's usually hard to regain any pulmonary function once you've lost it," she said. "To see mine go to that level is mindboggling."

Schaller has been able to stay out of the hospital so far this year whereas a few years ago, before she started exercising and participating in the clinical trial, it wasn't unusual for her to need inpatient care several times a year for three to five weeks at a time. She recently had a short-lived setback in her health and still coughs when she speaks but said this cough pales in comparison to the one she had before she started the new drug therapy.

"I feel incredible," she said. "Usually I'm in the hospital for treating lung infections with IVs three or four times a year, but I don't foresee myself needing that."

That may be a double blessing because Schaller isn't sure how much longer she can stay on her parents' health insurance, and she said she can't earn more than $900 a month without losing her Social Security disability payments. She runs the Rock CF Foundation from her parents' home in Trenton, Mich., but longs to live in a big city and become a

professional speaker.

"That's what keeps me up at night: insurance and income and living my dream, which I am (doing,) but I'd like to take it to the next step and live on my own."

Other adults may face similar challenges as more get diagnosed with cystic fibrosis for the first time.

People who are diagnosed as adults may have a delayed form of cystic fibrosis, but that doesn't mean it's necessarily a milder form, said Nick, who examined data on 156 patients diagnosed as adults and another 3,000 patients of all ages in the CF Foundation's registry over 15 years.

Of CF patients who had survived to age 40 no matter when they were diagnosed, about 85 percent eventually died of respiratory complications either before or after a lung transplant, he said.

Adults with suspicious symptoms may be referred to Nick's practice by primary-care doctors or specialists such as pulmonologists who've diagnosed them with chronic lung infection or assumed they had asthma or emphysema even though most patients never smoked, he said. Gastroenterologists may refer a patient with an inflamed pancreas if it's unexplained. Ear, nose and throat doctors may refer patients with severe sinus disease or chronic sinus infections, he said, and fertility clinics often send men who aren't producing sperm, another hallmark of the disease.

"No one is too old to have CF," Nick said.