

What doctors want patients to know about cystic fibrosis

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Cystic fibrosis (CF) is a lifelong genetic condition that primarily affects the lungs and digestive system, making everyday tasks like breathing and eating more difficult than most people ever realize or experience. Caused by changes in a single gene, cystic fibrosis leads to thick, sticky mucus that can clog airways, trap infection and interfere with the body's ability to absorb nutrients.

With newborn screening, earlier diagnosis and advancements in treatment, though, many people with cystic fibrosis are living longer, fuller lives than in decades past. But the disease still demands constant attention—from daily airway-clearance therapies to medications and clinic visits.

The AMA's What Doctors Want Patients to Know™ series gives physicians a platform to share what they want patients to understand about today's health care headlines.

In this installment, three physicians from University of Iowa Health Care took time to discuss what patients should know about cystic fibrosis. They are:

- [Anthony Fischer, MD, PhD](#), a pediatric pulmonologist and clinical associate professor of pediatrics and pulmonary medicine at Iowa Carver College of Medicine.
- [Douglas Hornick, MD](#), a pulmonologist and clinical professor of internal medicine-pulmonary, critical care and occupational medicine.
- [Michael Welsh, MD](#), a pulmonologist and professor of internal medicine-pulmonary, critical care and occupational medicine at Iowa Carver College of Medicine.

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Cystic fibrosis is a genetic disease

“Cystic fibrosis is an inherited disease caused by mutations in a gene called CFTR,” which stands for cystic fibrosis transmembrane conductance regulator, Dr. Welsh said, noting that everyone has two copies of the CFTR gene.

“If a person inherits one normal CFTR gene and one that is defective, the person will not develop cystic fibrosis, and they will never know they have one defective CFTR gene unless they are tested for it,” he added. “However, if both parents pass on a defective CFTR gene to their baby, the child will have two faulty CFTR genes and will have cystic fibrosis.”



Michael Welsh, MD

“Cells are surrounded by a membrane, and the CFTR gene makes a CFTR protein that forms a tiny channel or pore that allows the salt chloride to move across the cell membrane. In the lung, the salt chloride flows through CFTR channels into the trachea and bronchi,” Dr. Welsh explained. “This movement of salt draws water into the trachea and bronchi, helping mucus stay thin and liquid.”

“The mucus traps inhaled germs and dust so that the tiny hair-like cilia that are beating on the surface of bronchial cells sweep the mucus out of the lung,” he said. “When CFTR mutations produce a defective channel or no channel at all, the mucus becomes abnormally elastic and sticky, and the cilia cannot clear it out of the lung. The mucus builds up, clogs the bronchi, and the lungs become infected with bacteria and severely inflamed. The infection and inflammation will then destroy the lung.”

Symptoms of cystic fibrosis mainly affect lungs

“Cystic fibrosis symptoms mainly affect the lungs and digestive system,” said Dr. Welsh, noting that symptoms often include a persistent cough with thick mucus, wheezing, frequent infections, greasy or bulky stools, salty skin and poor weight gain despite hunger.

Other symptoms can include nasal polyps, delayed growth, fatigue and male infertility with severity varying by individual and age.

Diagnosis of cystic fibrosis starts with newborn screening

“It is caused by mutations in the CFTR gene and typically it’s diagnosed at birth by a newborn screen. That was a very important development,” said Dr. Fischer. “The newborn screen differs by state, but generally it works by first detecting a pancreatic molecule called trypsinogen.”



Anthony Fischer, MD, PhD

“Based on the level of immunoreactive trypsinogen that is in the newborn baby’s blood, that would lead to genetic testing of the same blood sample in the newborn to determine whether there are CFTR mutations as well,” he explained. “If a baby has elevated trypsinogen and has mutations in CFTR, then they may be suspected of having cystic fibrosis. They need to come in to get the gold standard test, which is sweat chloride measurement.”

“Babies who have a value of 60 or higher on the sweat chloride measurement or have two mutations in CFTR would be diagnosed with cystic fibrosis,” Dr. Fischer said. “The great thing about the newborn screen is that it also taught us a lot about CF. By applying this newborn screen in genetically diverse populations, we learn that cystic fibrosis affects people of pretty much every racial and ethnic background.”

Some people have one CFTR mutation

“We are also detecting people who have one CFTR mutation,” said Dr. Hornick.

“Traditionally I was taught that people who had one mutation, for example parents of patients with CF, didn’t have CF-related conditions.

“But now we know that a proportion do have CF-related conditions,” he added. “So, finding this out early in life may help identify that subset of individuals who could develop difficulties related to having one mutation.”

“Each biologic parent is going to be a heterozygote or a carrier, but most parents are healthy,” said Dr. Hornick, noting “there is some proportion who are carriers and have CF-related conditions, but what triggers that in that subgroup, we don’t know.”

Parents are at increased risk as carriers

“When I counsel new parents about this, I tell them there may be some increase in risk for you as carriers,” said Dr. Fischer. “You may be at higher risk than people in the rest of the population for things like pancreatitis, sinusitis or bronchitis.”

“I advise them to avoid the environmental risk factors that would add to that. For instance, if you smoke or drink heavily or if you forget to get your immunizations like the flu shot, that may increase your risk,” he explained. “Those are things that parents can do if they know that they’re a carrier.”

There are complications from CF

“The most common initial complication of cystic fibrosis is going to be pancreatic insufficiency,” said Dr. Fischer. “Intestinal symptoms are also common, and we don’t really understand the basis for them entirely, but many patients are going to have problems with constipation, diarrhea and abdominal pain.”

“So, we really need to have greater involvement of the gastrointestinal community in addressing those problems. In addition, CF related diabetes is an important complication, and we are always seeking input from the endocrine community,” he said.

Trikafta is an effective CF treatment

Elexacaftor-tezacaftor-ivacaftor (ETI) treatment, commonly known by the brand name of Trikafta, is a highly effective combination medication used for the treatment of cystic fibrosis. This medication is available for use in patients 2 years or older with at least one *F508del* mutation—which is the most common mutation causing cystic fibrosis—or other responsive CFTR gene mutations.

This treatment works by targeting the root cause of CF, fixing the defective CFTR protein to prevent thick mucus build-up in the lungs and the organs.

“It’s worth mentioning that ETI—the treatment for cystic fibrosis—remains inaccessible for people who have mutations that are not responsive,” Dr. Fischer said, noting “this often affects people who come from diverse ethnic backgrounds where *F508del* is not common.”

“This also affects babies because ETI is not yet approved for children younger than 2,” he said. “So, as a pediatrician, I worry more about my infants than I do about the teenagers because teenagers have an effective treatment. The babies don’t.”

Dr. Fischer also noted that, according to the Cystic Fibrosis Foundation, “after Trikafta came out, pulmonary exacerbations for younger kids, older kids and adults all dropped because of this treatment.”

A cystic fibrosis exacerbation is a sudden worsening of respiratory symptoms, such as increased cough, thicker or more sputum, and shortness of breath. These exacerbations can lead to decreased lung function, potentially requiring hospitalization and antibiotics.

New CF medications increase quality of life

“In 2012 is when the first drug became available based on Dr. Welsh’s research. It was called Ivacaftor or Kalydeco,” said Dr. Hornick. “It worked in 4% or 5% of the CF population, so it really had no major impact on the bulk of the CF population.”



Douglas Hornick, MD

“It wasn’t until 2015 when we had the first combination drug that helped move more functional CFTR to the cell surface in a larger proportion of the CF population. That started to have an impact,” he said. “Then we had another iteration of that in 2017 and then in 2019. At the end of 2019 was the drug that we currently use predominantly, which is available to 90% of patients with CF based on CFTR mutation status.”

“Before 2019, if you asked me how many inpatients I would have, I’d have half a dozen or more inpatients who I’d take care of. If you ask me now how many inpatients, it is zero,” Dr. Hornick said. “It’s had a huge impact on the quality of life of CF patients.”

Airway clearance is part of daily CF maintenance

When it comes to daily maintenance of cystic fibrosis for patients who are not candidates for ETI treatment, the key is “airway clearance, airway clearance and airway clearance,” Dr. Hornick said. This is so important to keep in mind.

Airway clearance techniques are daily, life-long therapies for cystic fibrosis that are designed to loosen and remove thick, sticky mucus from the lungs. This helps to reduce infections and maintain lung function. Airway clearance techniques include manual chest physical therapy, huff coughing, high-frequency vests for chests and breathing exercises.

“To give an example, I have a patient who has two mutations that are not responsive to ETI treatment, and we treat him with antibiotics, airway clearance and pancreatic enzymes,” said Dr. Fischer. “His lung function has steadily declined because he is not eligible for this medication.”

“For patients who are ineligible for ETI, treatments are very time intensive. It is similar to the era before ETI, when we expected that patients would perform 30 minutes of airway clearance twice daily in addition to inhaled therapies and pancreatic enzymes. It was a lot of work of just maintaining oneself,” he said. “And then when they got sick, they might have to come into the hospital for two weeks to treat an exacerbation.”

Families play a key role

“I’ve admired the way families that I’ve met over the years have adapted to managing this. They often do so creatively within their family unit,” said Dr. Hornick. “There’s an example in a video about Dr. Welsh’s work of a young woman who loved to run. Her way of dealing with it, which was an excellent way of doing airway clearance, was just running.”

“She would run and she competed at different levels early in her life and once she became an adult, it just was built in,” he added. “That’s just an example of some of the ways that patients and families will find the time and the effort.”

“I’ve taken care of patients with all kinds of lung diseases, but the CF patients and their families are somewhat unique and stand a little bit above the standard of what we see from patients,” Dr. Hornick said. “They seem to really take ownership for this and find remarkable ways to manage day to day and it’s admirable.”

“Most of these families have been really highly involved in assuring their nutrition and respiratory health from infancy,” said Dr. Fischer. “They would come in every single month in the first year of life, and then every three months after that just for regular checkups to assure that they’re doing OK.”

It's important to go to a cystic fibrosis center

“The [Cystic Fibrosis Foundation](#) deserves a lot of credit for this work. It’s a group that’s really advocated for children and adults with CF for decades,” said Dr. Fischer.

“In terms of what a patient with CF should know is, they need to go to a cystic fibrosis center,” said Dr. Welsh. “They need to have someone taking care of them who’s very knowledgeable about the disease.”

Patients can find a CF care center through the Cystic Fibrosis Foundation, along with other helpful resources.

More adults are presenting with cystic fibrosis

“Newborn screen, while it’s the most common way of making a diagnosis, it’s not perfect. There are regularly people who are missed by newborn screen,” said Dr. Fischer. “If people have symptoms suggestive of it, I refer them for testing.”

“We definitely hear a lot more about adults presenting with symptoms of cystic fibrosis who really have a long odyssey before they finally find Dr. Hornick,” he said.

For example, noted Dr. Hornick, a recent case was a patient who is about 26 years old who was referred to the liver clinic because he had cirrhosis.

“The liver doctor noticed he had a bad cough, which he downplayed. The patient had been downplaying or just seeing his primary doctor getting a course of antibiotics and then when he finally did pulmonary function tests, they were quite abnormal,” he explained. “That led him to be referred to a pulmonologist who then called me up. ... We did genetic testing and sure enough, he had CF and was diagnosed as an adult.”

“We’ve had a couple cases where an infant presents with a positive newborn screening and as we talk to the parents, they’ll start thinking, ‘Oh gosh, I’ve had a cough for a long time.’ Then we refer them to Dr. Hornick for testing,” said Dr. Fischer. “There have been multiple occasions where an infant diagnosed with cystic fibrosis results in an older person in the family—whether it’s a mom, a grandparent, an uncle or an aunt—also getting a diagnosis of cystic fibrosis.”

Eat a healthy diet

“When Trikafta came on board, our patients frequently were at the low end of the BMI scale—18 to 20 or lower BMI and we were trying to get them to 22 or 23,” said Dr. Hornick. “Now many of them have gained weight, they’re feeling so much better. The body’s energy usage for inflammation has gone down substantially.”

“We used to really push calories because it was impossible to get them to gain weight,” said Dr. Fischer.

With Trikafta, patients with cystic fibrosis are able to eat more and gain weight, but now the problems to keep in mind are obesity and diabetes, said Dr. Hornick, which is why it is important to eat a healthy, well-balanced diet.

Additionally, “exercise is so important too,” said Dr. Hornick.

Cystic fibrosis patients are living longer

“There are more adults than there are children with the disease because treatments have become much more effective,” said Dr. Hornick. “The adults are living longer, so they’re the bulk of the population.

“Before, when I first started, we were happy if they got to 20 years old,” he added. “Now the median survival of a child born with CF is 65.”