Jeanie Hanley

Cystic Fibrosis

I was diagnosed with Cystic Fibrosis late in life at 33, along with three of my eight siblings. In my childhood I was frequently diagnosed with sinusitis and bronchitis that always came back after antibiotic therapy. CF wasn't well recognized in people of color or different ethnicities, such as my Mexican-American heritage. I decided to become a doctor to find out what was wrong with my sister, Theresa, and me. She was the sickest of all of us.

Up to and throughout medical school, I would have sinus surgeries, two episodes of pneumonia and partially collapsed lungs (atelectasis) and too many courses of antibiotics. I was given the diagnosis of "atypical allergies and asthma" that was refractory to the usual asthma treatments except for oral steroids, which led to multiple courses, sometimes for months at a time.

During my medical training, I realized that the progressive respiratory issues plaguing my siblings and I was much more serious than just asthma. A microbiology class exercise where we coughed into an agar plate showed I grew the bacteria Pseudomonas. The professor took a look at the agar, a look at me, and deemed I looked fairly healthy so it must be a contaminant and not to worry about it. In another class, we had been learning about CF and how Pseudomonas is a common bacteria in CF lungs. Knowing my family's health issues, I did worry.

At my next doctor's visit, I mentioned this finding and asked if I could have CF. Unequivocally, he said no way – that I was too healthy-looking and too old, since the usual diagnosis occurs before seven years of age. He added that it only occurs in those with Northern European backgrounds, not in Mexican-Americans/Hispanic population. I learned I was the wrong ethnicity. He did diagnose me with "Medical Student Syndrome," found in medical students who think they have every disease they learn about.

In later training in Pediatrics and then in my Allergy subspecialty, I hoped other colleagues, allergists and pulmonologists could help me on my quest to figure out the underlying cause of my issues. Eventually I was diagnosed with ABPA for which I took more courses of steroids. However, it didn't explain my constellation of symptoms or those of my sister or other two siblings who also had recurrent respiratory infections and significant GI issues. Everyone I asked about CF said it wasn't possible for the same reasons I'd heard before.

When I didn't recover fully after a course of antibiotics, oral steroids, and daily asthma inhalers, I was told that I was being a "typical doctor" and not taking the medicines as prescribed. I was desperate to get better and did all that was prescribed in the hopes that it would help, but I was on my own.

After more episodes of atelectasis and hemoptysis, I had extensive workups to find another cause for the bronchiectasis that is common among people with CF, yet all tests and biopsies were negative. I had learned from my mother that one of my sisters had a negative sweat chloride test when she was a child. Being the gold standard for diagnosis for CF, it presented a sliver of doubt. I also learned that my brother discovered he had bilateral absence of the vas deferens (CBAVD) and couldn't have children. The mountain of evidence was growing when considering myself and my family's progressive symptoms, yet I still couldn't prove it.

In 1994, I attended an allergy conference and presented my family to a CF specialist (shout out to Richard Moss, MD, of Stanford) who encouraged me to obtain genetic testing, which had recently become available, on my entire family.

At Christmas time, I swabbed the buccal mucosa of all my siblings and asked a geneticist to order the testing, which checked for six CF mutations. The results showed that my mom, five sibs and I carried one F508delta mutation. I was very excited and presented the results to the geneticist. Yet he didn't bat an eyelash as he stated it was the normal carrier frequency in the community. He felt he was reassuring me by saying I was too old and too healthy-looking to have CF. It did give him pause, though, that the F508del mutation could occur in someone of Mexican-American descent.

Soon after, I came across a publication by a prominent researcher about two sisters diagnosed with CF as adults and whose sweat chloride was negative. I was incredulous and contacted the researcher. His research lab immediately offered to test the blood of my entire family including my parents. Again, at Christmas time, I drew all my siblings and parents' blood and sent them off. After two months, the lab confirmed that four of us had CF due to two CFTR mutations - F508delta and a rarer Class IV mutation.

Rather than being devastated, my sister Theresa and I were excited to start receiving the correct treatment and hopefully improve the quality of our lives. My other two siblings found the diagnosis harder to accept and didn't seek treatment at a CF Center for many years.

Only after this diagnosis would I learn that I could've been diagnosed with CF without genetic testing. Although my sweat chloride would be negative just like my sister's, another test for CF, usually performed at research centers like Stanford, called a "nasal potential difference" was found to be abnormal.

Unfortunately, the lifelong lack of proper treatment affected all four of my siblings and me. It led to more progressive bronchiectasis. Theresa had the worst disease and despite the appropriate CF therapies, she passed away eight years after being diagnosed from Cepacia syndrome – a CF complication and fatal bacterial lung infection that spread throughout her body. It's painful to think that an earlier diagnosis for her might have helped her live long enough to receive CFTR modulators that have helped me so much.

I also try not to predict what would have happened had I received proper treatment earlier. I was very sick during my allergy training, working full time, requiring constant antibiotics and steroids, nebulizing during commutes and at home, trying desperately to clear secretions. My CF has led to two bronchial artery embolization procedures to stem the flow of life-threatening hemoptysis, multiple sinus surgeries and yearly hospitalizations up until several years ago with the advent of CFTR modulators.

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My quality of life improved by leaps and bounds when I was fortunate enough to qualify for CFTR modulators, life-saving pills that jumpstart the mutated CFTR proteins and force them to function. They have improved my health to levels I hadn't known since I was a child. Even though I have to continue with all daily CF nebulizer and inhaled treatments to this day, my new level of health has enabled me to return to work as an allergist in private practice after over a decade on medical leave.

Spreading the word that CF does not discriminate and can occur in people of all colors, all ages, all countries and even in those with negative sweat chlorides is critical. Unfortunately, about 10 percent of all those with CF do not qualify for CFTR modulators due to having rarer mutations that modulators can't correct. Many are people of color and of non-European ethnicities, who may not be detected at birth by newborn screen. I suspect that the combination of my CF mutations would not have led to CF being diagnosed at birth by newborn screening. For this reason, I'm certain there are children and adults walking around with undiagnosed CF who will need the medical community's help to diagnose them.

Cystic Fibrosis

Cystic Fibrosis occurs when a person inherits a mutated (abnormal) copy of the CFTR (cystic fibrosis transmembrane conductance regulator gene) from each parent. It is an autosomal recessive disease meaning only people with two CFTR mutations have the disease. While there is no cure, life expectancy has steadily improved in the United States. Some other facts about Cystic Fibrosis are:

- There are now more adults than children with CF in the United States.
- Newborn screening for CF done on blood samples can identify most children before one month of age, which allows for early treatment and disease monitoring.
- CF individuals have abnormally thick mucus, which blocks the airways (obstruction) and leads to repeated infections and damaging inflammation in the lungs. Treatments are directed at trying to prevent and treat these problems.

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