Rebecca’s BHD Story
World Pneumothorax Day 2023

Hi, I'm Rebecca and I have Birt-Hogg-Dubé syndrome. I'm 41 years old. I reside in upstate New York in the United States. I'm a social worker in the acute adult inpatient psychiatric setting, and I'm mother to a seven year old little boy.

I began to experience symptoms of Birt-Hogg-Dubé in the summer of 2019, which started with some localized chest pain that wasn't resolving on its own and shortness of breath. After several visits to the emergency room with no answers, I then experienced my first pneumothorax in September of 2020.

During the Covid 19 pandemic, between September of 2020 and October of 2021, I experienced six pneumothorax with no reason why. In the fall of 2020, I began to experience worsening shortness of breath and more chest pain. I went to my local emergency room, very frustrated and upset after the advisement of my primary care doctor and was seen again in the emergency room. Every time I had presented to the emergency room with this chest pain and shortness of breath, my vitals were beautiful. My oxygen saturation was around 98 to 99%, and my heart rate looked great. The doctors were thinking that I had anxiety.

I began to experience the worsening shortness of breath with some sweating in fall of 2020, and that's when I had decided again to go back to the emergency room. This time when I went to the emergency room, I had also began to have an audible dripping sound in my chest. This was picked up by the emergency room doctor who recommended a CT scan. That was the first time I had received a CT scan in any of my emergency room visits, and this time it diagnosed me with a pneumothorax. The doctor came into the room and asked me how this could happen. He wanted to know if I had been in a car accident, if I had had any recent chest trauma, and I explained that I had not. After a few minutes, I kind of sat there for a second and wondered, could this be genetic? As my mom had told me that her lungs had collapsed when she was in her teens and her early twenties, and also that my grandfather, her father's lungs had collapsed on their own in the past. At that time, when the doctor re-entered the room, I did ask if this is something that could be genetic, and he didn't have an answer for me, but wanted me to see a pulmonologist.

I was admitted to the hospital on oxygen as I didn't need a chest tube at that time, and I began to, um, seek care under a local pulmonologist. The local pulmonologist looked at my CT scan and said that there were cysts in my lungs and it looked like I had emphysema. Although I had never smoked or vaped or done anything to compromise my lung tissue at this time. He said, you know, there's some diseases that women can have, but I don't think you have any of them. We're going to treat you with some oxygen and send you home.

I went home and three weeks later it happened again, except this time I did need a chest tube. This time it was much worse. This time I saw a cardiothoracic surgeon who looked over my CT scan and said, your lungs are full of cyst. I don't know why, but if we don't clear these cyst out, this is going to keep happening. I should take you to the operating room. Being that this was my first chest tube, I was terrified and I didn't want to go to the operating room. So he said, if it happens again, you're going to the operating room. Three weeks later, it happened again. When I had my first surgery, they operated on my left lung. I had a wedge resection with a mechanical pleurodesis. When they went in and

looked at my lung, the doctor said my lung was full of cysts and that he had to clean them out or my left lung was going to keep collapsing.

At that time, he didn't take a look at my right lung. He then sent the tissue out to a pathologist in the Rochester, New York area, and I was sent to see a pathologist and a pulmonologist and a geneticist in Rochester, New York at the University of Rochester Healthcare Center. At first, when I saw them, I had had to be done virtually. I had several virtual appointments where the doctor couldn't see my face up close and self-admitted that she wasn't very versed in the type of issues that I was having. She narrowed it down to two diseases, one being LAM and the other being Birt-Hogg-Dubé syndrome. When she first saw me after looking over the pathology reports and doing some questionnaires with me, she believed that I had LAM. This was devastating to me as the criteria to treat LAM, is much different than the criteria to treat Birt-Hogg-Dubé Syndrome. I was immediately started on the drug sirolimus and I was not having any improvement. This was in the summer of 2021.

After a couple months of being on sirolimus, my lung, my right lung collapsed in October of 2021. I went back to the same surgeon who had done my left lung and he said, you know what? We're not gonna mess around. We're gonna take you right back into the operating room. As I believe the same thing is happening with your right lung that has happened with your left lung. When they went in and operated on my right lung, it was much worse than my left lung. They removed about 15% of my lung, did another wedge resection with a mechanical assist, and then tacking my lungs to my diaphragm and my chest wall lining to keep them inflated so I wouldn't keep experiencing this.

At this time, I was sent back to University of Rochester Healthcare Center to meet with a different pulmonologist who was more versed in what was going on with my lungs. So at this time, I was finally able to see a pulmonologist face to face and they were able to see me without my mask on, and they were able to see the skin on my face, and they were obviously seeing fibrofolliculomas, which they were not able to see over the camera.

While I was at home, I met with a geneticist in the University of Rochester Healthcare System and I was tested for the FLCN gene, which came back positive in January of 2022, and at that time, I received my concrete diagnosis of Birt-Hogg-Dubé Syndrome.

I believe that raising awareness for Birt-Hogg-Dubé syndrome is important because it affects both men and women. It is a disease of the kidneys, the skin, and the lungs. In my case, I had six pneumothorax before I was able to get a diagnosis and I had presented to the emergency room several times to be sent home possibly with a mild pneumothorax and a diagnosis of anxiety. With the help from the Birt-Hogg-Dubé Foundation and the National Registry that was created this past year and Facebook groups, I've been able to find support with other folks who've been diagnosed as well.

My hope is that doctors will take spontaneous pneumothorax more seriously and realize that there could be a genetic cause to it, and that it's not always trauma based. My advice for those who experienced a genetic cause of a pneumothorax or are still waiting for answers is to not give up and to keep seeking treatment throughout the United States and the rest of the world. There are pulmonologists who are quite versed in Birt-Hogg-Dubé Syndrome as well as Nephrologists and dermatologists. My hope is that with the recent creation of the registry, as well as the Facebook groups, that we can find support for a disease that has no cure and is little known.