Relapsing Polychondritis Is a Lung Disease: My Story

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Relapsing polychondritis is a rare autoimmune disease that can be fatal. This systemic condition with a predilection for cartilage can inflame the trachea, distal airways, ear and nose, blood vessels, eyes, kidneys, and brain. If relapsing polychondritis is not diagnosed early and treated effectively, there can be irreversible damage to the cartilage that can lead to tracheal or bronchial collapse, blindness, and deafness.

I am training to be a physician/scientist, and I have severe relapsing polychondritis with prominent involvement of my airways. Here is my story (Figure 1).

Figure 1. The author, Marcela A. Ferrada, M.D.

Unraveling the Mystery

I knew something was wrong with me, but I did not know what.

I am trained in critical care and infectious diseases. My husband is a cardiologist. My sister, brother-in-law, and father are surgeons. My two best friends are intensivists. None of us could come up with a diagnosis.

Now, looking back, I realize that I had intermittent symptoms for many years, including several episodes of arthralgias, tendonitis, sinusitis, dizziness, syncopal episodes, severe fatigue, voice changes, and knee swelling. When I felt bad enough to see a physician and get blood work done, all appeared normal. The results were reassuring but, in retrospect, dangerous, because they led me and my doctors to believe that my symptoms were inconsequential and perhaps even psychosomatic. Sadly, this is what many people with my disease experience, feeling that they are imagining things, until it is too late to prevent highly consequential organ damage.

I became much more concerned about my health when I developed shortness of breath and a persistent, nonproductive cough about 8 months before I was diagnosed. I also had anterior neck pain, dysphonia, and aphonia. The coughing was sometimes so severe that I broke a rib. I was diagnosed with asthma, which seemed to be confirmed when I responded to a short course of prednisone.

I felt better after taking the prednisone but continued to experience shortness of

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This is my personal story. The views expressed are my own and do not represent the medical research institution where I am employed.

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breath on exertion, which became progressively more debilitating. The day I completed my fellowship training in infectious diseases, I was particularly short of breath. I titrated my prednisone dose to control my respiratory symptoms. However, every time I lowered the dose, I experienced pleuritic chest pain, shortness of breath, cough, costochondritis, and pain and swelling of my hands and knees.

While I was suppressing my symptoms with prednisone, not knowing what was wrong, I served for the first time as an attending physician in the intensive care unit. I had been dreaming about this advancement for years. To control my symptoms during long, demanding work days, I took more prednisone—no one knew how much. I was determined not to allow my disease—whatever it was—to get in the way of that special time in my life. But I knew that something was seriously wrong. Whatever it was, I feared that it would truncate my dream of becoming a career intensivist.

Shortly after I completed my first attending rotation in the intensive care unit, I consulted an amazing rheumatologist. On August 6, 2015, he told me that I had relapsing polychondritis. I learned that my shortness of breath and my relentless, breath-taking, energy-draining cough were likely caused by laryngeal involvement. Later on, I also learned that the shortness of breath was possibly due to inflammation in my small airways (a little-known manifestation of relapsing polychondritis) (Table 1).

I was so relieved to have a knowledgeable and empathic doctor. For the first time, I felt safe. We implemented a targeted treatment strategy. Initially, I required up to 170 mg of prednisone a day to quell my inflammation. Taking a high dose of a corticosteroid is terrible! We as doctors sometimes underestimate how distressing the side effects caused by these drugs can be, such as the hump that forms at the base of the neck. If you ever have a patient with a prominent hump, do advise him or her to buy a specially shaped pillow—it will make a huge difference.

Every day during the initial phase of controlling my disease, while I suffered with pain and breathlessness, I thanked God for being alive. For 3 weeks, I thought that each new day would be my last. Every morning, I awoke with a sensation that someone was cutting my throat with a knife. I never thought I could endure such pain, and I really was not sure that I would ever get better.

Controlling Flares

On a novel immunomodulatory regimen, I was able to wean off prednisone after about 9 months. However, about a year later, I contracted influenza from my 3-year-old daughter. Lowering the doses of my immunosuppressive agents for just 3 days to help me fight off the infection triggered a severe flare of my relapsing polychondritis involving my inner and outer ears, throat, nose, multiple joints, airways, and lungs. By modifying my immune-modulating regimen, we were able to regain control of the disease, but unfortunately this regimen became less and less effective over time.

To reduce my susceptibility to flares, we are now trying medications that likely have never been used in relapsing polychondritis before. I can say happily that sometimes I feel like a laboratory animal. I feel proud and lucky to have the opportunity to do this, to know that I may be able to provide treatment options that work for other patients with relapsing polychondritis.

Over the past 2 years, I have recognized that in my particular case certain foods can also trigger flares, including wheat, beef, and soy. Because there is no clear evidence of the association between food and disease activity, I tried hard to believe that these were not triggers. Regrettably, I proved myself wrong several times. It is hard to have a disease that does not allow you to do what you want to do or eat what you like to eat. I also learned that running can trigger a flare. I used to run half marathons before I got sick. It is difficult to adjust to, recognize, and accept your physical limitations, but every time I realize that I am having thoughts of self-pity, I remind myself that I can breathe, I can walk, I can work—things that I was not able to do for months before.

Understanding Relapsing Polychondritis

I am fascinated and intrigued by relapsing polychondritis. I think about the disease day and night. I started reading about the disease as soon as I was diagnosed.

Since then, I have not been able to stop reading and rereading the literature. I have learned that the disease is exceptionally difficult to diagnose. There are no known disease-specific biomarkers, and all of the usual, nonspecific inflammatory markers can be completely normal, despite significant tissue inflammation. Biopsy, when positive, can be helpful but can also trigger a flare. By default, diagnosis is based entirely on clinical-radiographic-pathological correlation and remains imprecise. Clinical diagnostic criteria formulated in the 1970s have not yet been updated, and currently there are no guidelines for classification or management (1, 2).

Relapsing polychondritis is indeed a "rare disease" (one to four cases per million per year) (3). I also suspect that relapsing polychondritis is underdiagnosed, because too few physicians think or even know about it. Even more importantly, I think it is a "forgotten disease."

We just forgot about it! I heard someone once say that she thought it was a

Table 1. Pulmonary function test

Date	FEV L (% predicted)	FEV_1/FVC L (% predicted)	TLC L (% predicted)	RV/TLC % (% predicted)	FEV_1 pre-BD L (% predicted)	FEV_1 post-BD L (% predicted)
July 8, 2015	2.6 (85)	96 (111)	4.3 (96)	45 (145)	2.6 (85)	1.71 (64)
July 29, 2015	1.7 (66)	83 (97)	4.2 (94)	50 (159)	1.75 (66)	1.23 (46)

Definition of abbreviations: BD = bronchodilator; FEV_1 = forced expiratory volume in 1 second; FVC = forced vital capacity; RV = residual volume; TLC = total lung capacity.

"dead-end disease," basically not worth studying at all. I disagree. First of all, I believe that all diseases are worth studying. Second (and of course my opinion is self-serving), relapsing polychondritis is a fascinating disease, with so much potential for discovery that may ultimately improve the lives of patients suffering from this illness.

Physicians who know a little about relapsing polychondritis tend to think of it as a rheumatic condition that can cause peculiar inflammation of the outer ears. Relapsing polychondritis is also very much a respiratory and cardiovascular disease, and respiratory manifestations are the most common cause of death.

Relapsing polychondritis can affect the nasal passages, larynx, trachea, and larger airways, focally or continuously. Inflammation destroys the structural integrity of the cartilaginous rings of the larger airways, causing luminal collapse during expiration (tracheo- and/or bronchomalacia). Apposition of the two surfaces of a fish-mouthed airway may trigger a sensation that gives rise to paroxysmal, nonproductive coughing. In turn, forceful coughing can cause tight collapse of malacic segments of the central airway, cutting off expiration prematurely. Airway injury can also predispose to recurrent or chronic bacterial infection. In addition, I believe that relapsing polychondritis can affect the distal airways, causing radiographic changes that are not yet well characterized in the literature (4).

Patients with relapsing polychondritis may present primarily with respiratory symptoms, including nasal and sinus symptoms, dysphonia, cough, and shortness of breath. Some of the symptoms may be episodic. Particularly when onset of the disease leads with respiratory symptoms, an erroneous diagnosis of asthma is often made and retained, even when symptoms do not respond to ordinary treatment for asthma. Relapsing polychondritis is not a silent disease, but you have to know and think about it to make the diagnosis.

My Personal Commitment

More than 90 years have elapsed since the first case of relapsing polychondritis was described in the medical literature (5), yet we still know so little about the cause, manifestations, natural history, and treatment of the disease. I decided to address our ignorance head on. I accepted that my health would not allow me to excel as an infectious disease and critical care specialist and that my chosen specialty would not enable me to focus on helping patients with relapsing polychondritis, so I enrolled in a rheumatology fellowship.

I want patients with relapsing polychondritis to be as functional as possible, to have the opportunity to receive treatments that work for the disease, and to be able to have a life like I do.

I am extremely fortunate to work in an amazing and supportive environment. Together with outstanding scientists and clinical investigators, I am working to more completely describe the clinical manifestations and natural history of relapsing polychondritis and to classify subtypes. I am aiming also to unravel the mechanism of the disease and possible genetic associations to improve diagnosis and treatment.

Reflections on Living with a Rare Disease

After I acquired my diagnosis, when I was very sick, I often turned to an online support group for information and solace. As difficult as this has been for me, I have benefited from management by an expert clinician and by extraordinary support. It breaks my heart to know that many others suffering from this disease are not as fortunate.

As a patient and physician advocate for those afflicted by relapsing polychondritis, I believe that clinician awareness is the first key to improving the diagnosis and care of patients who suffer from the disease. I urge pulmonologists to learn the clinical manifestations of this rare but devastating respiratory disease and to consult a rheumatology colleague when faced with a symptom complex that may be explained by relapsing polychondritis. Asking for help is a sign not of weakness, but of wisdom.

Realizing that I can die at any time has changed how I experience my life and how I set my personal goals. I believe that every day should count, and there is no time to waste. I have become more compassionate, tolerant, understanding, and happy. I am no longer physically able to fulfill my earlier dream of excelling as an intensivist, but I have taken on a new goal: to be of lasting service to people who are afflicted with a rare, forgotten disease. I will not rest until I achieve my new goals.

Where there is research there is hope, and we have research. ■

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