In the past twelve months, my pursuit of a well-rounded understanding of life within the rare disease community led me on a powerful three part journey: an honest introspective exploration of the role of disease in my life, an emotional new relationship with a caregiver, and a deliberate examination of the interaction between the disease community and our society.

This story must begin within myself, specifically on the seventh chromosome of every cell in my body, where an error in the DNA for a small transmembrane protein has managed to dictate much of my life. This one amino acid exchange is responsible for my diagnosis of cystic fibrosis, one of nearly seven thousand rare diseases that exist today. When I say that this faulty gene has dictated much of my life, I do not say it with resentment, because it has steered me in many positive directions. Reflecting on my personal path to medical school, it is clear that I was drawn to the medical field because medicine is so intimately woven into my daily life, and I know that I will be a better physician because of the education that can only come from being a lifelong patient. I suppose the most essential lesson that I can glean from my experience with disease and carry into my career is the penetrating truth that patients can live full and meaningful lives despite the intrusions of chronic disease.

The funny thing about living with a disease that is present from birth is that it does not begin as feeling unique. In fact, it feels perfectly ordinary, the only normal that you know. I have never known a life where I did not need to swallow a handful of pills before a meal or inhale nebulized medication throughout the day. However, in elementary school, I was hit with the reality of how foreign my lifestyle is when I gave a short presentation on cystic fibrosis to my class. I'll never forget the isolation that I felt when, on the playground later that day, a classmate said, "You shouldn't even come to school because you're just going to die soon!"

Admittedly, the message is harsh in the blunt words of a seven-year-old, but there is a spark of truth in the theme that could have persuaded my parents to limit my world. The line between false hope and hope may be paper thin, but herein lies the empowering magic of living with rare disease. My parents gave me the hope of a fulfilling life, constantly supporting me to reach above and beyond expectations. As supporters of

patients with rare diseases often do, my parents encouraged me to disregard the unsubstantiated limitations, and because of this, I was able to strengthen a foundation of knowledge that would support me for a future career. This is a topic that I have heard again and again through patient stories: after removing our confining limitations, we will see patients rise above and beyond expectations through amazing adaptive capabilities. I see such a beauty in the empowered patients and caregivers; a beauty that I know it is my goal to support through a career of research, care, and advocacy. The strength of hope is truly powerful. This is the first of many defining lessons I appreciate when I look back on my life as a patient.

My reflection made it clear that hope is a great motivator and also brought to light an immensely important aspect of life for the rare disease patient: relationships. Just as my growth was dependent on the support of my parents, many rare disease patients rely on caregivers. From my own life, I can appreciate this from the patient perspective. I know firsthand how being prescribed another "5 minute" medication, while seemingly convenient from the physician's point of view, might require big sacrifices from both caregiver and patient. As a child, I observed those prescriptions spark lengthy phone calls with insurance companies for my mother, a frustrating necessity that I appreciate so much more now that I have struggled with those calls myself. Additionally, there can be the equipment preparation, sterilization, and clean-up, perhaps in the chaotic setting of siblings that also require attention from the caregiver. But this framework begs another question; What is the experience like for the caregiver?

This is the question that launched part two of my journey. My personal reflection left me intrigued by the influence of disease on family dynamics and its potential to cause huge shifts in values and priorities. I know cystic fibrosis enabled my family to bind together to beat it, but I wanted to know what it feels like for someone who joins the fight voluntarily. Please note that the names of the individuals involved in this element of my growth have been changed where appropriate to protect their privacy.

The relationship that shaped my understanding of the caregiver perspective began at a chance encounter at medicine grand rounds. It was only the second month of my medical training, but the assignments and pressures were already starting to pile sky-

high, leaving what felt like no room at all for anything that was not mandatory. However, on this day, the description of the guest speaker caught my attention. Incredibly, Rachel was bravely advocating for the rare disease community by speaking about her experiences as a caregiver for a man with cystic fibrosis. I entered the lecture hall hoping to gain some insight into the caregiver's perspective and left having truly gained a new lifelong friend and mentor.

When she was just seventeen, Rachel fell in love with a boy named Chris, a boy who happened to have cystic fibrosis. At that time, she learned that he was expected to live only until the age of thirty or so, but, when you are young and relatively healthy, that prognosis can haunt you daily and yet seem so profoundly distant. I know the feeling all too well, but Rachel gave me a gift of a perspective that I have not lived. My eyes welled with tears as I listened to her simultaneously joyous and heartrending true story of a young couple approaching mortality and the life that they squeezed out of their ten years together. I was entirely engrossed in this opportunity to peer into the thoughts of a caregiver. In addition to learning how the disease affected their relationship, I got to hear her reactions to her late husband's lung collapse and frequent doctor visits, but that was just the beginning of Rachel's impact on my life. After the hour was over, I was invigorated, inspired, and more determined than ever to help the rare disease community.

I was completely overcome by a flood of emotions when I first spoke with Rachel. Our common experiences with the threats of cystic fibrosis ignited a conversation that spilled into our next few meetings. Our conversations have made it clear to me that fighting a rare disease forces the creation of an extraordinary sense of purpose and a fierce drive to advocate for awareness and improved outcomes. Every time we communicate the momentum builds, and I am still invigorated with each exchange because she reminds me that the community that I am seeking to help is full of strong and incredibly worthy individuals.

I am so grateful for the perspective that she has shared with me over the past year. In her years by Chris's side, Rachel observed more than enough patient-physician interactions to critically consider the aspects of a good doctor. When asked what she thinks the most

important characteristics are, Rachel recalls the last week of Chris's life. It was nearly two years after his double-lung transplant, and a serious infection was proliferating in his lungs. He was put on a ventilator, and Rachel sat nervously by his side, fruitlessly brewing over all worst case scenarios. As a doctor in training, I can imagine myself as the physician looking in on this difficult situation and sympathize with the temptation to offer hope to the grieving spouse. Yet, this is where that delicate line between hope and false hope requires precise differentiation, and I am so grateful that Rachel was able to open up to me about this incredibly intimate, vulnerable moment so that I can see the importance of this distinction. The unfortunate truth was that the odds that Chris would live were very slim. Fighting the lure of providing false hope, Chris's doctor held Rachel's hand and told her the truth with compassion. This conversation allowed Rachel to be with her husband fully in his last days, and she feels that it was only possible because the physician had made the effort to know her well enough to know she would want to know. This sensitive conversation shaped Rachel's relationship to Chris's passing and highlights the significance of not only the patient-physician relationship, but the caregiver-physician relationship as well.

My conversations with Rachel have taught me that, as a physician, it is acceptable and even preferable to never pretend that medicine has all of the answers, but to be openly and deeply engaged in the mystery. She had the most respect for honest physicians who would be open about the fact that some treatments were experimental, as is often the case with rare diseases. Rachel feels strongly that aside from providing necessary medical treatment, the most important role of a physician is to understand the patient as a whole person apart from their illness. Caring for an individual with a rare disease doesn't simply begin and end with the patient. Many patients rely on the support of a caregiver, so, generally, the patient's health is linked to that of the caregiver. Therefore it is vital to pay attention to the health of the patient as well as their support system. My relationship with Rachel has given me the invaluable gift of a greater awareness of the personal struggles and joys of caregivers.

Sharing stories with Rachel unveiled an appetite for hearing more personal experiences, not just within the cystic fibrosis community, but with other rare diseases. I started consuming stories wherever I could get my hands on them: podcasts, documentaries, or

in the hospital. Each new story makes it more apparent to me: this is the patient population for which I am entering the field of medicine. I want to be the physician that helps these families feel supported and hopeful. I want to find a treatment that will prevent someone like Rachel from seeing their loved one pass away because of one faulty gene.

I decided that I wanted to find some way to support children with rare diseases as a medical student and started searching for something that I could be involved in. I came across an organization called Positive Exposure, founded by an award winning fashion photographer, Rick Guidotti. On a break from a photo shoot one day, Rick happened to see a girl with pale skin and white hair, a girl with albinism. He was stunned by her beauty, and when he returned home, he looked up albinism. He was upset to see that the images he found were sad and dehumanizing. Energized with the desire to change the way we see disease, he stopped working in the fashion industry and created a not-for-profit organization. This organization seeks to spread positive images of children with genetic diseases, so that they can "be seen as their parents see them, as their friends see them, as valuable and positive parts of society, as beautiful." Rick's energy is contagious, and I cannot wait to bring it to my medical school to allow other students to follow his call to action. I believe that by spreading beautiful images of children with differences, we can begin to spread awareness and build empathy among physicians and future physicians. Additionally, the time of initial diagnosis can be particularly frightening, especially if an internet search of the disease reveals dehumanizing photographs. I hope that Positive Exposure can instead offer hope in that period through positive media that highlights the beauty of the human beings rather than magnifying the fear caused by a disease.

Through Rick, I was introduced to another brave individual living with a rare disease, Kafi. At birth, Kafi was abandoned by her mother because she was born with albinism. In the superstitious part of Africa in which Kafi was born, there is a belief that the limbs of a person with albinism can bring you luck in life. Kafi told me that she was frequently called "money" by strangers on the street because her body parts could be sold for profit to witch doctors. Tragically, Kafi was brutally attacked and lost her leg because of the prevailing belief that her body parts can be used to bring luck. Her circumstances force

her to live with a fear that I am grateful to have never known, but her story has a common thread with many of the other rare disease stories I have heard. Living with a rare disease can lead to isolation because of an appalling lack of awareness. Kafi is a heroic advocate working to spread the knowledge of genetics to help her countrymen understand the cause of albinism and protect those with albinism in Africa. I truly believe that through fearless advocates willing to share their stories and the brilliant work of crusaders like Rick Guidotti, we can cultivate understanding and combat this paucity of awareness.

The culmination of inspiring new relationships and lessons of this formative year of personal and professional growth sparked the idea for an event that I am hosting for International Rare Disease Day this February at the University of California, San Francisco. My dream for this event is to enhance awareness for rare diseases in general, provide an uplifting day for patients and families, and to strengthen interest among medical students in the field of rare diseases. The bold act of advocacy that Rachel took on by sharing her personal story with a room full of strangers was incredibly inspiring to me. Reflecting on how our relationship changed me, I am struck by the power of personal stories and the way they enrich understanding and foster empathy. Certainly such a powerful tool could be used to inspire the next generation of physicians. To harness this power, I look forward to having three patients with rare diseases and one mother of a child with a rare disease tell their stories and answer questions at our Rare Disease Day event. These individuals will be a valuable educational resource for us by allowing us into their world through storytelling. Additionally, Rick Guidotti will be capturing his stunning images of children in our community with rare diseases, and we will create an exhibition from the shoot. I feel that helping patients and families cope with illness has a lot to do with dispelling isolation, and I hope that this event will provide an empowering atmosphere for building a sense of community. This has been a powerful year of discovery for me, and I look forward to watching this year become the foundation of my career as a physician in the rare disease community.