The Importance of Rare Disease Education

Sophia A. Walker
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Recently a wise professor told my class that we medical professionals are some of the most powerful people in the world. Indeed, we have the ability to meet people at their most vulnerable, sometimes on the very worst day of their lives, and help them. “This profession,” he told us, “is such a privilege that we must never miss the opportunity to have at least done some good for every patient.”

Over the past several weeks, as I have planned rare disease awareness events and begun preparing to enter the clinical years of my medical education, I find myself considering these words more frequently. However, at the end of the day, I wonder how powerful we are, really...

My interest in rare diseases originated during my senior year of high school, when I first started volunteering at the National Organization for Rare Disorders, Inc. (NORD). I was overwhelmed to discover the many obstacles experienced by patients who have rare diseases. On a technical level, any disease that affects fewer than 200,000 Americans is considered rare. Of the more than 7000 rare diseases, only approximately 350 have treatments that are approved by the US Food and Drug Administration (FDA). I found that individuals with these diseases, almost two thirds of whom are children, show great courage and perseverance in the face of significant discouragement. Although I had always wanted to be a doctor, it was not until I witnessed such unrelenting determination to overcome barriers in healthcare that I discovered my own enthusiasm for medicine.

All physicians strive to provide attentive medical care with the utmost compassion and empathy; however, as medical professionals, we must also be our patients’ most vocal advocates. Although I was not yet a physician, I still wanted to contribute to this effort. I wanted to provide a forum for the nearly 30 million Americans with rare diseases whose voices often go unheard in the medical community, and I wanted to share this passion with my peers. Every year, my fellow students and I host a Rare Diseases Awareness Event. Patients, students, clinicians, and researchers come together to share their experiences and insights regarding rare diseases. We strive to shed light on the lives of these individuals by allowing them to share their own stories, sometimes for the very first time.

Although many students may assume that we do not need to know as much about rare diseases because we are unlikely to encounter them in our practice, this is simply not the case. In fact, every one of us preparing for medical careers will see patients with rare diseases, and the extent to which we prepare ourselves for this reality will determine the impact we can have on these patients’ lives. Patients who have
A rare disease face difficulty in every step of medical care, including diagnosis, treatment, and preserving quality of life. Sometimes, patients go years without receiving the correct diagnosis for their condition. Once they finally have an answer, often no treatment is available for their condition. As future physicians, we must aim to improve these prospects; the first step in doing so involves developing a keen understanding of this patient population.

The opportunities for medical students to learn about rare diseases are vast. Gaining a basic understanding of how the experience of having a rare disease is different from having a more common disease is equally essential. The National Institutes of Health (NIH) has great information related to rare diseases on its website, and the NORD website provides overviews and links to more than 200 patient organizations that provide excellent information about specific rare diseases. Students can also apply for a free NORD student membership by writing to bhollister@rarediseases.org. Once you register, you receive a monthly eNews and quarterly newsletter specifically designed for students planning healthcare careers. If you're attending the American Medical Student Association annual convention in Washington, DC, on February 27 and 28, come to the NORD booth in the exhibit hall where patients with rare diseases will be sharing their stories.

With each speaker I listen to at a rare diseases event, with each new person I meet, I am filled once again with immense pride that our efforts, if even in a small way, have done some good. Unlike many people who are involved in advocacy efforts in this area, when I began this work, I did not have a personal connection to rare diseases. However, after years of getting to know people who have experienced these struggles, I can say that I now have several. In fact, it is the memory of the individuals I have met and the satisfaction in having contributed to raising awareness that has guided my interests, served as an influence in many decisions, and ultimately has been the driving motivation in achieving my aspirations. With every step I take moving forward in my career, rare diseases comes along with me and will continue to do so.

My passion for rare diseases advocacy has become perhaps the foremost aspect that defines me and has made me who I am. It has given me direction, has made me a leader, and continually prepares me to become one of those physicians who will do some good. A couple of years ago, one of my undergraduate professors asked me, "Are you that rare diseases girl?" He went on to say that a student who had been inspired to research rare diseases after attending my event had approached him with an interest in working in his lab. This is the reason why I raise awareness for rare diseases. If just one more person every year becomes inspired, that may eventually make all the difference in the world. It turns out that, in the end, we are all powerful together. After all, according to the NORD motto: "Alone we are rare. Together we are strong."