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Inspiring Compassion for Rare Diseases

Colton Margus

When I was asked to contribute to this volume of *Quill & Scope*, I was thrilled to have the opportunity to introduce Student Advocates for Neglected Diseases (STAND), the new student group that I started here at New York Medical College to spur greater student engagement with this small and most in-need patient community.

Despite what we learn in the classroom, many medical students across the country are not sufficiently exposed to and thus not fully appreciative of the deeper burdens that patients and families with rare and neglected diseases face. I want STAND to address that, because I believe doing so will lead to more informed and compassionate physicians.

Rare diseases are often life-threatening or chronically debilitating, and each rare disease affects a very small portion of the population (fewer than 200,000 people in the United States).¹ Neglected or orphan diseases include both rare inherited disorders as well as neglected infectious diseases affecting the global poor. Such diseases are traditionally not given a high priority for prevention or treatment and, as a result, lack sufficient attention from governments, healthcare professionals, news media, and drug developers.

The Orphan Drug Act of 1983 and the subsequent Rare Diseases Act of 2002 exemplify laudable efforts to “promote the development of drugs and devices for rare diseases” through establishing a national office, increasing research funding, and improving incentives for development of new therapies.¹ However, the complexities of rare and neglected disease diagnosis at the level of individual patients and families are still underappreciated within the medical profession.

At the heart of the matter is the belief that all patients should matter. Neglected conditions have a harder time attracting funding and support, and patients often suffer from that oversight. Tens of millions of Americans are affected by nearly seven thousand rare diseases, most of which lack any treatment or cure. Their need for a helping hand does not depend on disease prevalence—and neither should our compassion.

What should be great news for busy medical professionals and students is that the smallest effort for the smallest communities can often make the biggest difference. Having an impact on a rare disease does not necessarily mean finding a cure. Coping with a rare disease can be incredibly isolating, and an outstretched hand from the medical community can mean the world. Just spending one-to-one time with the patients and their families and showing genuine interest in what life is like for them can provide tremendous encouragement. It can alleviate their feelings of being forgotten and provide encouragement that the next generation of physicians will be sensitive to their needs.

Even beyond the orphan communities themselves, progress on behalf of neglected diseases helps to advance the medical field more broadly. A substantial part of today’s medical knowledge originated in the pursuit of an overlooked question or with a rare disease model.² Future physicians must appreciate that neglected diseases can offer fertile opportunities to advance innovative new research strategies and treatment methods for more common disorders. In fact, as healthcare management shifts toward personalized medicine targeting smaller and smaller subsets of the general population,³ one could argue that our research models developed on behalf of rare disease communities will be all the more relevant.

I am admittedly biased—two of my younger brothers suffer from a rare genetic disease called ataxia telangiectasia (A-T). At a birth frequency of 1 in 300,000, my brothers’ autosomal recessive truncation of the ataxia telangiectasia mutated (ATM) gene is extremely rare, but no less devastating in its stunted growth, progeric aging, cancer predisposition, immune deficiency, and cerebellar degeneration.⁴ On top of coping with their sons’ diminishing quality of life, my parents have had to fight indifferent insurance companies and navigate enormous bureaucracies for support. I have seen doctors misdiagnose and misguide treatment, but I have also seen wonderful physicians listen carefully to my family’s concerns and recognize the importance of collaborating with patients and their families in managing a disease. I have seen the enormous difference a good physician can make to families like mine, even when there is no cure.

Now that I am in medical school myself, I want to take the opportunity to convince receptive future doctors of the value in championing and understanding rare diseases just as they begin their careers. I hope to plant the seed in the next generation of healthcare professionals by getting medical students to think about and to engage with patient communities that they might never come across otherwise.

STAND is busy organizing several events on campus to bring attention to rare disease patients and their families. For example, following our introduction to the DNA-repair disorder xeroderma pigmentosum (XP) in our biochemistry course last spring, we invited Caren Mahar, patient mother and founder of the XP Society and Camp Sundown, to visit New York Medical College. It was a great opportunity for students to hear a more personal perspective on the disorder, and to better understand the often overlooked burdens on families diagnosed with incurable diseases they cannot even pronounce.

Still, as much as lectures and presentations raise awareness, nothing compares to the effect that direct, individually-driven interactions can have on one’s outlook, investment, and



propensity for future engagement. That is why STAND is brainstorming more creative ways to get the student body directly involved with patients, both on campus and at other medical institutions.

Our newly created “David R. Cox Prize for Rare Compassion” will recognize the essays of student doctors who have most inspiringly engaged a rare or neglected patient community that they have encountered. Participating students will meet with an unfamiliar patient, family, or advocate affected by a rare disease, and share their newfound insights afterwards in a short essay. Beyond that, it is also my hope that with help from the Global Genes Project, an international organization, we will be able to facilitate Rare Disease Day activities at medical schools around the country. There are a lot of exciting directions we can go in.

Throughout this process, it has been overwhelming to see how many people have come out of the woodwork for something meaningful. Even in the group’s infancy, we have already garnered the support of students, administrators, and faculty alike, and their collective response has been more than I could have hoped for.

I feel incredibly fortunate and look forward to seeing the integration of our classroom foundations with rare disease advocacy and patient engagement. I am optimistic that our doing so will make a positive difference in our community and in how at least one doctor approaches patients and their families somewhere down the road.

Author’s note: Associate Dean Tony Sozzo’s support has been invaluable in the success of this student group, and a meeting in January 2014 organized by Dr. Jennifer Koestler reiterated the school’s commitment not only to supporting student initiatives but also to adapting the New York Medical College curriculum and learning experience in a way that will produce even better doctors.

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