Thoughts From a Medical Student: Helping Future Physicians Understand Rare Diseases

By Colton Margus

Soft drink and fast food companies often make children the primary targets of their advertising. These companies know that people frequently establish consumption habits at an impressionable young age that extend well into adulthood. Such a long-term strategy isn’t just smart business. It is essential to the survival of the brand.

As a second-year medical student with two younger brothers who have a rare genetic disease – ataxia telangiectasia – I think the rare disease community can learn and benefit from the strategies of these corporations.

Just as these companies target future consumers before they are actually customers, we in the rare disease community should be making a stronger effort to reach future physicians before they are practicing medicine.

Last year, I started an advocacy group among medical students called Student Advocates for Neglected Diseases (STAND) and established the David R. Cox Prize for Rare Compassion. Both initiatives are aimed at inspiring future medical professionals to become more aware of, and identify with, rare and neglected disease communities.

Medical students actually learn a great deal about orphan diseases in our coursework already, but that education is too often limited to the corresponding mutations and common clinical presentations tested on licensing exams. We have far less exposure to the people behind the ailments – the children and families waiting desperately for a diagnosis and treatment.

Raising awareness of rare diseases, while important, isn’t enough. The rare disease community needs to inspire future physicians to become champions for our patients. We want doctors not only to “get it” but to want to be a part of it, as well. They have to become personally committed to improving the lives of rare disease patients and their families.

The benefits of having competent young physicians coming into their careers not only understanding rare diseases but thinking critically about them are numerous. Young physicians bring new insights and fresh perspective that challenge old treatment regimens and notions. They also add critical support through case reports on unique clinical presentations, natural history studies, and even the development of patient registries that will facilitate future clinical trials and make rare diseases more attractive targets for drug development down the road.

The stories of rare disease patients are inspiring and motivating. Often, all that is necessary to convert compassionate bystanders into active supporters is simply to provide first-hand glimpses into the daily struggles faced by patients and their families. The dramatic impact that rare disease medical experts can have on the lives of their patients may also be very appealing to those entering the medical professions.

If you (like me) are a medical student, please feel free to get in touch. And if you (like me) are affected by a rare disease in one way or another, please write to rarediseaseday@rarediseases.org to ask how you can get involved in NORD’s outreach to physicians and other medical professionals.

Colton Margus is a second-year medical student at New York Medical College.
Responses to Thoughts From a Medical Student: Helping Future Physicians Understand the Rare Disease Experience

Jo says: January 22, 2015 at 11:22 am

Bravo Colton! You are right on target with this initiative and I wish you all the best with it. As a rare disease patient myself and a full-time advocate, I have attended a fantastic program for 2nd year med students every March for the last 8 years or so. It aims to introduce them early on in their course to a variety of patients, in my case, the rare genetic disorder, MEN2a. I talk to a room full of students about my experiences, hopes, fears and answer any questions they care to throw at me. Evidently they find this incredibly useful. I just wish more medical educational institutions would run such programs. Many patients are more than happy to get involved in this way. Good luck with STAND!

Mommy Outside says: January 22, 2015 at 2:29 pm

Yes to this! We have a daughter with an arguably “rare” condition and the lack of knowledge out there is only matched by the lack of desire to obtain that knowledge. It became increasingly clear through our experience that even the medical professionals that had any understanding of the condition were almost bullied into not pursuing it because of the lack of support from other professionals. It’s really sad and it’s really scary when you are left to make a diagnosis yourself and hope to hell you can find a doctor who is willing to step out of the box and treat it.

Karen McEwen says: January 22, 2015 at 2:44 pm

Wonderful article Colton! I have an 11 year old daughter that suffers from a rare disease called Primary Ciliary Dyskinesia (PCD). To raise awareness about this life altering disease, we have done 2 local tv interviews and had a couple of articles written in our local papers. I would love to be able to go to colleges to speak and educate med students about PCD but I have no idea how to get started. Any tips you can send me???? Thanks!

Kim McIlney says: January 22, 2015 at 4:41 pm

As a family physician no longer practicing because of multiple sclerosis, with a daughter with an extremely rare disease, I APPLAUD you for taking the initiative to do this! While we learn about diseases in medical school, the focus is on the medical therapy to implement, not on what life is really like... or how to “treat” the significant life-changes that these diseases cause. PLEASE feel free to contact me if you want to discuss further or if I can be of assistance to your group!

Deb O says: January 27, 2015 at 3:05 pm

Thank YOU for your comments! It must be incredibly challenging for you to be on the “other side of the fence”. We go home after a doctor appt, and don’t know what to do next. There’s no one to guide us about what action to take in a rare disease that has no treatment. Doctors are quick to recommend hospice for diseases with low quality of life, but how does providing $140/day (medicare) to an organization really help the patient manage the complexities of a rare disease? There are MORE of us with rare diseases (combined) than AIDS AND Cancer! 1 in 10 What medical professionals fail to see on those fancy power point slides of the patient, PCC, SP, PT, OT, Neuro, social worker, etc, is that the CARE-GIVER is ALL ENCOMPASSING and they usually aren’t even listed on it! We MUST support a different approach by doctors for those with rare diseases!

Deb O says: January 27, 2015 at 2:47 pm

I am in awe of your inspiring message! You are 100% correct that just raising awareness ISN’T ENOUGH. My mom is facing a losing battle with Progressive Supranuclear Palsy or Corticobasal Degeneration. My impression is that doctors must get more involved with the patients and that spending 1.5hrs every 3-4 mos is NOT enough to absorb an accurate picture of what is happening to the patient. It’s so easy to understand the science of it on a molecular level, but far harder to understand the variety of manifestations of the disease on the patient, especially when the patient is “atypical” for the rare disease. And if not being able to offer a treatment makes the dr feel powerless, imagine how it feels to the patient!!! We are told to “have hope” when there is no hope for progressive conditions. Doctors should not fear getting to know the patients beyond their office, because that is where the patient LIVES their disease. Family caregivers are not trained in “medical” symptom identification and may not be relaying imp info. And doctors should not fear advocating for “off label” or “compassionate” use of drugs/therapies for persons whose time is limited. Aren’t you becoming a doctor to help? Helping beyond the office setting is critical. Thank you, Colton Margus, for doing above and beyond to inspire the future medical professionals!

As the Director of Medical and Professional Programs for a rare disease non-profit organization, Cure HHT, I would be interested in partnering with STAND to educate medical students about HHT (also known as Osler-Weber-Rendu Syndrome). We agree completely that educating younger generations of patients and physicians is crucial to better diagnosis, treatment and care. I look forward to speaking with you.

Amanda says: January 23, 2015 at 10:06 am

Thank you, thank you, and thank you! I am glad that to hear that future doctors want to learn and educate themselves on multiple rare conditions. My son was just diagnosed with Chronic Granulomatosus Disease and basically immune compromised. The whole ER staff look at me like I am crazy and I made up this condition. Not only that it brings down my anxiety as a parent knowing my children are in the right hands and understand how to proceed. I just want to say good luck and keep doing what you are doing!

S. Smith says: January 26, 2015 at 6:00 pm

So glad to hear this coming from a medical student–I totally agree! It took 9 years and seeing numerous physicians before one finally knew what was wrong with my husband–NMO. Finally, a diagnosis. Most people don’t understand the frustration of going through something like that. Thanks for your efforts.

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