EDITORIAL

Using rare diseases as teaching models to increase awareness

The institutionalized definition of a rare/orphan disease varies, depending on geographic location. A disease is considered "rare" if it afflicts fewer than 200,000 people in the United States, less than 250,000 people in the European Union countries, and fewer than 50,000 patients in Japan.[1] It is noteworthy and sobering to learn that for many patients with a rare disease, appropriate diagnosis often occurs more than 5 years after the onset of signs and symptoms.[2] Moreover, almost half of all such patients are initially given the wrong diagnosis, and a subset of these patients has a history of 3 or more incorrect diagnoses before the correct diagnosis is finally rendered.[2] On average, patients with rare diseases visit at least 7 different physicians before receiving an accurate diagnosis.

Because there are over 7000 recognized rare diseases and millions of people live with these diseases, encountering such a patient in clinical practice is not rare.[1] To this end, as in the case of many readers of *Oral Surgery, Oral Medicine, Oral Pathology, and Oral Radiology*, I have directly contributed to the diagnosis and care of several patients with rare diseases. In some instances, the diagnoses I rendered provided, for the first time, some of these patients or their caregivers with an actual name of a disease to explain their years-long history of unexplained signs and symptoms. For some individuals, the emotional impact of simply obtaining an actual diagnosis and then further discovering that there are support groups for people who are similarly affected cannot be underestimated. Patients afflicted with rare diseases face significant health disparities and delays in diagnosis because their physicians are often not sufficiently informed.[3] Thus, finding a community clinical practitioner who actually knows something about the disease may also be profound for some of these patients.

Studies have shown that knowledge of rare diseases among health care students is poor but that targeted educational approaches may be an effective means to increase awareness.[4,5] As an educator, I have always been frustrated when students ask, "Why do we have to learn about these rare diseases that we are never going to encounter in clinical practice?" I was recently asked this question, but as a sign of the times, it went one step further. A student stated that if he ever did encounter a patient with a disease that he "never heard of" he would "Google it" to learn more about the disease. I felt I should at least acknowledge that he would be interested enough to investigate the details of his future patients' disorders. However, the student further predicted that he would very likely refer the patient to another clinician because he thought he would never feel comfortable treating the patient himself. Admittedly, I had to take a deep breath and collect my thoughts to ensure I delivered a measured response.

In conversations with colleagues engaged in dental and medical school teaching, an overwhelming sentiment I have encountered is that the current generation of students is "different" from those in previous years. No single explanation is given to clarify why they are perceived as "different." Their reliance on immediate information gleaned from social media is often suggested as a possible reason. However, this "give me only what I need to know" mentality is not a new phenomenon.

The first time I was asked the "Why do we have to learn ..." question, I was in my first year as a faculty member and a freshly minted oral pathologist. I distinctly recall responding defensively by stating, "You're going to be a dentist in a couple of years. Everything you learn in this class is important!" Yet, as I was saying it, in my mind, I knew that my answer was the equivalent of the classic, childhood non-answer: "Just because!"

Fortunately, over the years, my response to this question has matured because my understanding of science and of patient care continues to evolve. I have come to realize that using rare diseases as teaching models can be quite effective as a basis for learning fundamental principles of basic science and clinical practice. The important, added bonus is that the students simultaneously learn about a rare disease that they may not have the opportunity to learn about elsewhere. Moreover, I have had a number of students express interest in engaging in basic science research projects after discussion of rare diseases presented in lecture.

We must collectively strive to continue to increase awareness of rare diseases. As an educator, pathologist, and clinician-scientist, I firmly believe that this must begin at the level of predoctoral dental and medical education. In my opinion, the "need to know" does not have to be mutually exclusive from the so-called "nice to know." I believe that educating students on rare diseases is a win-win for everybody involved—students and patients alike.

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https://doi.org/10.1016/j.oooo.2019.04.015

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