Improving Rare Disease Recognition Through Undergraduate Medical Education

By Muhammad Shaikh – University of Leicester

Runner-up in the Student Voice Prize 2019

An essay competition hosted by Findacure and Medics 4 Rare Diseases

A rare disease is defined by the European Organization for Rare Diseases as a disease with a prevalence of less than 0.05%[i]. However, the World Health Organisation estimates that as many as 1 in 15 people may be affected by a rare disease[ii]. Rare diseases are rare, but *patients* with rare diseases are not. Hence, it is vital that doctors are adequately equipped to manage these patients.

Patients with rare diseases see many doctors and may be misdiagnosed multiple times before their condition is fully understood and diagnosed. This is what is known as the diagnostic odyssey. In some senses, this is an unavoidable problem. Medical schools train students in pattern recognition as it relates to pathology[iii] (regardless of the style of teaching on the didactic versus experiential spectrum). Students learn to associate certain signs and symptoms with specific diseases. Because teaching time and resources are limited, exposure to – and familiarity with – common disease patterns is prioritised. This focus comes at the expense of familiarity with rarer conditions. Students also learn – and rightly so – to prefer more common differential diagnoses, as they are statistically more likely.

This is particularly evident when preparing for examinations – the keystone of the medical student's existence. Here the advice is often to focus specifically on having an encyclopaedic knowledge of the handful of most commonly diagnosed conditions in each speciality, as opposed to having a broad knowledge of the speciality. This adequately prepares the student for the vast majority of patient encounters, and safeguards them against gross malpractice.

Subsequently, once the student becomes a doctor, the problem is compounded. It is only natural that clinicians' knowledge of the conditions they face will be directly related to prevalence. Knowledge of conditions encountered more often will be reinforced, whereas knowledge relating to conditions encountered less often will be slowly but surely forgotten.

This not to argue for inaction, but rather to understand the diagnostic odyssey as an artefact of the way doctors are trained. With this in mind, how can medical education be enhanced such that doctors are better equipped to recognise and manage rare conditions? To answer this question, I will focus on measures that can be implemented in the standard five-year MBChB course, as this is most relevant to my current situation and experience; I do not wish to speak beyond the limits of my knowledge.

Familiarising students with each and every rare disease – of which there are thousands[iv] – would be practically impossible. The rate at which medical knowledge is increasing is meteoric. (The rate at which the medical student memory is increasing is – sadly – not meteoric.) In 2020 it is projected that the interval required for the doubling of medical knowledge will be just 73 days. Students who graduate in 2020 will have experienced four doublings in knowledge.[v] Medical

knowledge is increasing at a rate that outstrips our abilities of assimilation and application. Adding more content to medical courses and increasing their length is simply not a viable strategy. Rather, we need to adopt a training method based more on principles and organisation of knowledge.

Perhaps one measure which addresses the problem in this way would be to present the 'rare disease patient' to medical students as a unique profile, much in the same way students learn about the polypharmacy patient with their many medications, or the complex patient with their numerous co-morbidities. Students should have direct contact with rare disease patients, ideally following their systems-based teaching, to allow for a full medical apprehension. Direct patient contact would help students develop a comprehensive understanding of the patient within the biopsychosocial model, and allow them to grasp the difficulties of the diagnostic odyssey. Students should be introduced to the diagnostic pathway for this type of patient – why to take a detailed family history, how and when to refer to clinical genetics services (80% of rare diseases are genetic in origin[vi]), and what additional support the patient may need. Students should also be acquainted with the resources available for the management of rare diseases, such as online encyclopaedias and orphan drug directories. Being a doctor in the modern day is not about knowing all medical knowledge. Rather it is about two things: knowing the scope of medical knowledge, and knowing the sources of medical knowledge. (This, apparently, was not the explanation my parents were looking for when questioning my grades.)

One thing which should not be omitted in this teaching is the necessity of empowering the patient to actively partake in their own medical care. All patients should be empowered: the best medicine is prevention, and preventative medicine involves enacting things only the patient has control over. However, rare disease patients arguably benefit more from taking control of their own care than any other group. These patients often know much more about their condition than the clinicians treating them. However, even without a diagnosis, patients can be very astute when it comes to researching their potential conditions – after all, no one knows a patient's symptoms better than the patient themselves. Additionally, the empowerment of rare disease patients has led to political action[vii] which has ensured more investment into this patient group from the medical and pharmaceutical industries[viii].

This teaching should also be reflected in medical school examinations, with questions specific to rare diseases. Rather than directly assessing rare disease knowledge, which would be somewhat unfair at early stages of education, the students' diagnostic reasoning, procedure, and management could instead be assessed. Desirable outcomes on an assessment of this kind would include the ability to exclude more common conditions and the knowledge of what avenues to pursue when suspecting a rare disease.

Moreover, students should be exhorted to be mindful of the humanistic aspect of care. Focusing on the disease rather than the patient can have negative care outcomes[ix]. Sometimes the best treatment that can be offered as a doctor is empathy and understanding.

Another measure could also be to directly teach students some of the more common 'rare diseases' (such as Huntingdon's disease) in relation to their systems teaching, to allow them to

more rapidly recognise them in clinical practice. This need not come at the expense of other course content; every course has inefficiencies that could be removed to make room for more teaching time. The changes need not be drastic: even encountering a disease once during the course and never hearing about it again could be enough for students to consider a rare disease diagnosis in a patient, which may mean the world for them.

Discussions regarding these diseases could also take place during clinical rotations and specialty training – students may often find this more engaging than reviewing the most common diseases ad nauseum.

A measure which medical schools may also consider relates to the recognition and treatment of rare diseases, but has further scope also. This measure would be to include in the syllabus a student-led module based on medical research, with the eventual goal of publication of a short paper by all students. This would have a number of favourable outcomes, including potentially contributing towards students' Foundation Program Application System (FPAS) points. However, the most significant outcome would be increasing students' familiarity with the scientific process and with medical literature. The importance of this cannot be overstated; it is not just the case that the medical journals are the first recourse of the clinician suspecting a rare disease, but it will be through these journals that they may discover many rare diseases in the first place. After all, the doctor is a lifelong learner.

A more light-hearted measure – which is suggested entirely in earnest – is to encourage students to watch medical dramas in their free time. Many, such as *House MD,* or *Hannibal,* feature rare medical conditions prominently, and present them in an interesting and memorable way. The production teams often work with medics to ensure medical conditions are portrayed accurately. Some may scoff at this being a serious suggestion, but when medical schools do little preparation of their students to encounter rare medical conditions, some action is better than none at all.

The quandary of the diagnostic odyssey is an unfortunate one. Its existence is an artefact of medical teaching, and a natural consequence of the prevalence of rare diseases. Nonetheless, medical schools should prepare students for encountering rare diseases in a much more effective manner, as rare disease patients are often encountered. I have made a number of suggestions to this end, while recognising the limitations of time and resources medical schools find themselves under.

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