If you hear hooves it may be a zebra

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Medicine tells us that ‘common things are common’. While this is an obvious truism, it is estimated that 3.5 million people in the UK and 200 million people worldwide suffer from a rare disease. Therefore, no matter what specialty you belong to, it is very likely you will come across patients with rare diseases.

Although rare diseases are not strictly a medical category in that they are not linked by a common pathology or set of symptoms, they are grouped together because they share a plethora of problems. These include diagnostic delay, limited research, poor treatment options and lack of awareness among the medical community.

The Council of the European Union formally acknowledged the need for an organised approach towards tackling these issues when the recommendation was made that each Member State develops a rare disease strategy by the end of 2013. The Department of Health managed to get there just in the nick of time with the UK Strategy for Rare Diseases announced in November 2013.

One of the areas this report highlighted was the need to improve time to diagnosis through ‘better training on rare diseases on university courses (undergraduate and graduate)’.¹ A group of medical students at Barts and the London School of Medicine and Dentistry has teamed up with a patient advocacy group (The UK Lysosomal Storage Disorders Patient Collaborative Group) to form the organisation Students4RareDiseases to try and raise awareness about the importance of considering rarer diseases. As future clinicians, we believe that it is important medical students appreciate that the term ‘rare’ is not synonymous with irrelevant.

This idea often results from a misconception about what the term rare actually means. In Europe, rare diseases are classified as those seen in 1 person out of 2000 of the population or less. Of the 6800 rare diseases, some will be very, very rare, while other conditions are more common such as cystic fibrosis.

Students4RareDiseases aims to bring medical students into contact with clinicians and patients

at educational talks and events so that they have greater exposure to rare diseases. The key message at these events is not to be an expert in all rare conditions but to take away some useful tips that could help to diagnose a rare disease in the future.

At the annual Rare Diseases Symposium, which was held at the Royal Society of Medicine this year and featured six talks by a patient and five leading clinicians, the overwhelming majority of students reported that the speakers were instructive and met their educational needs, particularly in highlighting the importance of considering rarer diseases in the differential diagnosis. The event proved to be so popular that we have been invited back next year, confirming that there is demand for this kind of education among medical students and doctors.

Nearly half of patients with a rare diseases report receiving an incorrect diagnosis, with one-third of these receiving more than three before eventually being diagnosed correctly.² This delay not only leads to poorer clinical outcomes but also presents considerable psychological distress to the sufferers and their families. Barriers to timely diagnosis include lack of awareness among the medical community, symptoms that overlap with common diseases, and lack of defining clinical hallmarks and simple diagnostic investigations. These features undoubtedly make the task of timely and accurate diagnosis difficult. We believe that the solution however is relatively simple. The basic tools of medicine – history and examination – should be used to formulate a relevant list of differentials which, while of course including the more common, should also include the rare. In order to do this in the first place though, medical professionals need to be alert to the possibility of rare diseases.

If differential diagnoses are packed with the rare then ruling each one out becomes a laborious task that is not only costly in terms of time and resources but also causes undue stress for the patient. The point, however, is just to include the conditions that fit with the clinical picture (which is what we all should be doing anyway).

What is particularly frustrating for many patients is that although they may exhibit the classic symptoms that are typical of their rare disease, it is assumed that they are suffering from something much more common. This happens even if the more common cannot account for the clinical picture; contrary to the popular phrase, it looks like a ruff, swims like a ruff and squawks like a ruff, but still gets called a duck. A great deal of time is lost due to this failure of inductive reasoning.

Of course it is unrealistic to expect doctors to recognise every single rare disease, or even the majority of them. This message is echoed in the Department of Health's strategy which stresses the need to ensure 'all doctors are alert to the possibility of rare disease when they see patients, even if they are not able to diagnose specific diseases'.¹ After all, medicine has been categorised as a system of specialties, each containing doctors with knowledge finely tuned to a particular field. This makes the task of diagnosing a rare disease easier: identify the main system that appears to be affected and then refer to the relevant specialist doctor as is done for any disease. The real skill in this process is knowing which patients to refer on, particularly in general practice, where ever-squeezed budgets further complicate this decision. The fact of the matter is that at present most patients with rare diseases report a struggle to access their specialty service, and every incorrect diagnosis will ultimately carry its own significant financial burden.

Ultimately, as a collective, rare diseases are common and to manage this considerable group of patients we need to ensure timely diagnosis in order to deliver effective and appropriate care. The crucial gateway to diagnosis is having informed and responsive doctors who use the evidence in front of them, and not just the textbook highlights.

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