

When You Hear Hoofbeats

In a concise reflection, consider a specific aspect from RDNZ's White Papers that intrigued and/or surprised you. Explore the implications of this aspect, emphasising how it has the potential to illuminate your career path in health. Discuss how this knowledge about people living with rare disorders will contribute to the way in which you will "show up" for people living with a rare disorder in your professional life as a healthcare practitioner, i.e. how it will shape your future approach to patient care and commitment to rare disorders.

'When you hear hoofbeats, think horses, not zebras.' How many times have I heard this adage, while standing red-cheeked following admonishment by a senior doctor? 'Common things are common' can be an efficient - if occasionally reductive - diagnostic algorithm, useful for medical students in their early years. However, when this mental model continues to be broadly applied in clinical practice, we risk prematurely narrowing down possible diagnoses without sufficient consideration or investigation. After all, in a hospital serving a large enough population, 'rare' can be seen every day.

While Aotearoa New Zealand has no official definition for a rare disorder, the European Union defines it as affecting less than 1 in 2000 people. In Aotearoa, up to 6% of the population has a 'rare' disorder, equating to approximately 300,000 people. RDNZ's White Papers contain a number of key insights for those clinicians that encounter rare disorders in their practice. When we consider the figures above, we can appreciate this means everyone.

Two aspects of RDNZ's White Papers stood out to me as particularly surprising. Firstly, the lack of continuity of care (with 69% of respondents having to visit three or more doctors to get a diagnosis) and the alarming rate of misdiagnosis (with three in ten being misdiagnosed at least twice). This indicates that there is a flaw in how many doctors approach 'unusual' presentations, causing patients to spend valuable time, money and energy, bouncing around the system. Secondly, coding for rare disorders not being routinely incorporated in health systems datasets means that this percentage of our patients go uncounted, and unconsidered in planning and budgeting. This is a systemic error of the healthcare system that urgently requires addressing. In medical school, we use the word 'heterogenous' to describe diseases or disorders that are the result of several different root causes. Rare disorders are heterogenous in nature, but, as this report shows, it is often the effect of engaging with a healthcare system ill-designed to cater to their needs, that results in similar outcomes. To address these issues, we need to continue to upskill healthcare providers not necessarily in *what* they know, but to be dedicated to ongoing learning and growth, and adept at reassuring patients in the face of diagnostic uncertainty. We also need to ensure our system is adequately equipped for accurate data collection for better planning and service delivery.

Sufferers of rare disorders live in a world of hypotheticals. A paucity of evidence means even the predictions of experts contain a great deal of uncertainty. However, the act of

walking alongside a patient through their journey, viewing them first and foremost as a person, and secondly as a patient, can be a humanising experience which can itself be healing. In the career I will soon embark on I hope I am brave enough to advocate for policy change where it is required, humble enough to admit when I am wrong, and open to learning with and through my patients, in all the years that lie ahead.

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