Syndromes and patterns

How to make sense of it all?

Stephen A Margolis

> *Nature uses only the longest threads to weave her patterns, so that each small piece of her fabric reveals the organization of the entire tapestry.*
>
> – Richard P Feynman[1]

I vividly remember the challenge of learning how to allocate seemingly disparate pieces of clinical information into a composite whole. Having what I thought at the time was a reasonable grasp of basic physiology, I still struggled to sort through the seemingly endless lists of potential items in the history and physical examination and place them in priority order. Why in this circumstance was it essential to focus on this specific aspect of the medical history, while that was not the case for the next person? And then, how to make sense of the information?

I have always been interested in how we perceive patterns from seemingly incongruent pieces of information. Why is the 'answer' outwardly self-evident after being found, yet invisible beforehand? What is the sentinel event that triggers the shift in one's thinking? And how does this occur?

One particularly helpful underlying theoretical understanding is to consider Kuhn's paradigm shift, where development of understanding follows a discontinuous rather than a linear path of smooth progression, punctuated by sudden insights arising from thinking laterally and beyond the boundaries.[2] For example, if eczema is very common in my practice due to local context, simple probability theory predicts that most new rashes seen are likely to be eczema. So why would I consider that this rash is in fact a different diagnosis while contemplating the person in front of me? Perhaps the trigger to consider important, yet uncommon, diagnoses is when there is one or more disparate symptoms or signs; that barely discernible yet nagging voice inside that keeps saying, 'There is something not 100% right here, so let's move laterally'.

Unfortunately, in some instances the pattern does not fit any specific pathological-based disease diagnosis and you are left with the syndrome – a characteristic combination without clearly defined pathophysiology. What is required are scientific advances to move from a collection of signs and symptoms (syndrome) to the focused (specific pathophysiological disease). A telling illustration is non-specific chest pain. During my early years while in forensic pathology, I commonly saw sudden unexpected death due to coronary artery disease (CAD) in those recently started on treatment for syndromic upper abdominal symptoms. In retrospect, these people had misdiagnosed angina, consequent upon the limited understanding and investigations of that era. With the advent of readily accessible, reliable and safe investigations, we now routinely diagnose CAD even when the presentation is more obtuse, providing the opportunity to institute appropriate, specific care. Progress in diagnostic tools has siphoned off syndromic presentations into disease, giving rise to the prospect of more effective and targeted treatments.

The corollary is managing patients whose presentation has been compartmentalised to a specific syndrome, where on further lateral reflection a specific disease is more likely. Human nature seemingly struggles to consider alternatives once labels have been applied. Perhaps the best examples are those considered to have chronic pain or fatigue whose management may focus on the psychological. Advances in diagnostic processes and understanding may later demonstrate specific pathophysiological processes that are amenable to adding new, focused treatment in addition to the psychological. Or, with additional time, the underlying disease process becomes more advanced, visible and responsive to standard diagnostic processes. The challenge in each of these circumstances is knowing when to re-evaluate one's thinking.

As knowledge evolves, the scientific implications of findings in history and examination change. People presenting with undifferentiated symptoms and signs require both linear and lateral thinking for accurate diagnosis and hence correctly targeted treatment.

Author

Stephen A Margolis OAM, Editor-in-Chief, *Australian Journal of General Practice*; Professor, School of Medicine, Griffith University, Qld

References

1. BrainyQuote. Richard P. Feynman quotes. Mercer Island, WA: BrainyQuote, [date unknown]. Available at www.brainyquote.com/quotes/richard_p_feynman_160463 [Accessed 9 May 2022].
2. Kuhn T. The structure of scientific revolutions: 50th anniversary edition. 1st edn. Chicago, IL: University of Chicago Press, 2012.

correspondence **ajgp@racgp.org.au**