

The gaps in specialists' diagnoses

Specialists need broad expertise in diagnosing clinical problems arising from diseases involving different organ systems

On average, about 10% of primary care visits result in a referral to a specialist,¹ and of these, up to half relate to diagnostic uncertainty.² Diagnostic error is estimated to occur in between 10% and 15% of clinical encounters.³ Medicolegal concerns loom large around missed or delayed diagnosis of potentially serious conditions such as heart disease or cancer. Patients often present with non-specific symptoms and signs, especially in the early stages of emerging illness, which can be accentuated in the complex context of multiple comorbidities, frailty or other disabilities. Accordingly, a broad differential diagnosis that includes diseases of more than one organ system has to be considered, followed by a recursive refinement of diagnostic probability in the face of uncertainty.

Limitations of specialist expertise

When confronted with such challenging scenarios, clinical expertise restricted to single organ systems may be a limitation in formulating differential diagnoses. Evidence suggests that specialists practising outside their areas of expertise are less efficient diagnosticians and provide lower quality care than generalists, even after adjustment for illness severity.⁴ When it comes to diagnosing routine cases, specialists practising within their (single domain) specialty tend to use a highly specific, data-driven (forward) approach, in which diagnoses are generated from data by applying a small set of if/then production rules based on prior, well organised knowledge. Intermediate hypotheses tend not to be generated or evaluated.⁵ When dealing with complex patients, this expert prior knowledge is automatically activated such that what an expert already knows influences how they perceive and solve diagnostic problems. When confronted with non-routine or atypical cases, specialists tend to generate more diagnostic hypotheses relevant to their specialty domain, and assign these hypotheses higher probabilities, compared with hypotheses outside their domain that may be just as, or more, salient.^{6,7}

Clinicians of all persuasions are often more anchored to their initial diagnostic formulations, and more resistant to changing them, than they should be in light of conflicting objective evidence.⁸ Other forms of cognitive bias such as confirmation bias, premature closure and framing effects add to the problem.⁹ Vignette studies suggest that clinicians' level of confidence in their diagnoses bears little relation to either diagnostic accuracy or degree of case difficulty.¹⁰ Indeed, the mismatch between diagnostic accuracy and confidence appears to be worse for more difficult cases.¹⁰ This increases the risk of diagnostic error, which may be compounded by failure to fully elicit contextual factors — psychosocial, financial and cultural determinants of health — that influence the mode of presentation and add more complexity in formulating a correct diagnosis.¹¹ Further, when diagnostic errors made

by one doctor are recognised by other doctors who subsequently see the patient, such errors are rarely communicated back to the original doctor for fear of endangering professional relationships.¹² An opportunity to engage in reflective practice and foster trust is therefore lost.

Patients with different comorbidities may present with problems reflecting disease—disease, drug—disease or drug—drug interactions. Clinicians trained as generalists seem better at navigating this complex mixture of issues than single content domain specialists.¹³ Lack of experience in diagnosing and managing such patients may explain more cross-referrals to other specialists, longer stays and higher resource use incurred by single domain specialists compared with generalists within hospital settings.^{14,15} It may also explain why patients with specific chronic diseases managed by such specialists are often undertreated for other disorders unrelated to their primary condition, disorders that may ultimately assume greater clinical importance.¹⁶

If complex cases remain unrecognised as such and hence undiagnosed, single domain specialists may consult other specialists on the basis of a “best guess” as to which organ system is likely to be incriminated; for example, “I think this is a nerve problem and you need to see a neurologist. It does not appear to be a joint or muscle problem which is my area of expertise”. The extent to which selection of specialists for cross-specialty referrals is appropriate given the clinical scenario is uncertain, although one study suggested that generalists select more appropriately than specialists, perhaps reflecting their arguably broader social networks or range of clinical acumen.¹⁷ Over-referral driven by diagnostic uncertainty can come at a cost of fragmented care “by committee”, unnecessary investigations, potentially inappropriate polypharmacy, increased costs and more patient confusion and isolation as a result of being caught in a “pass the parcel” sequence of referrals. Each consecutive consultation carries the risk of each different specialist overestimating the predictive value of symptoms and signs as portending serious conditions related to their specialty, which may lead to overordering and overinterpretation of diagnostic tests. Specialists may be driven to overinvestigate because of an intolerance of anxiety and uncertainty within their domain of expertise, compounded by fear of reputational loss, or worse, from missing that “one in 1000” (rare) chance of a specialty-related diagnosis. However, the increasing availability of more sensitive diagnostic tests may simply breed more uncertainty by generating more false positive results or disclosing unrelated and mostly benign lesions (known as incidentalomas).¹⁸ While an exhaustive list of specialty-specific investigations may find no abnormality within a particular organ system, this may not provide diagnostic closure for the patient and may generate frustration and loss of trust in the system of care as a whole. In an audit of 120 specialist reply letters to general practitioners for

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referrals that involved diagnostic issues, only 69 (57.5%) stated a specific diagnostic formulation.¹⁹

Enhancing diagnostic skills outside expertise domains

Several options are open to specialists when faced with diagnostic challenges outside their area of expertise. First, they could look to upgrade their generalist skills, although achieving this could be problematic because of limited time available or credentialing requirements that mandate exclusive focus on one specialty. Given that specialists now outnumber GPs in Australia, there may need to be a commensurate step back from (over) specialisation to generalism, in recognition of the fact that diagnostic acumen in the face of increasing complexity is a specialisation in its own right.²⁰ Becoming more self-sufficient in diagnosing unfamiliar presentations involving growing numbers of older patients with multimorbidity may require more in-depth training and continuing professional development in general internal medicine,²¹ and a commitment to lifelong learning that includes breadth as well as depth. Such a need is being recognised by the significant proportion of advanced physician trainees in adult medicine in 2017 undertaking dual training (ie, training in another specialty in addition to their core specialty [436/2840; 15.4%]), of whom two-thirds (286/436; 65.6%) are undertaking training in general medicine (Linda Smith, Chief Executive Officer, Royal Australasian College of Physicians, Australia, personal communication, 2017).

Second, in perplexing cases, specialists might seek the (early) advice of generalists skilled in generating and prioritising differential diagnoses and determining the most parsimonious selection and sequence of investigations for arriving at a final diagnosis.²² Third, a conscious effort should be made to reconcile diagnostic formulations for a new problem with the list of pre-existing problems in discerning whether the former represents an atypical presentation of an organ system disease outside the (single) domain expertise of the treating specialist.

Fourth, there needs to be more emphasis on improving diagnostic calibration (ie, a match between hypothesised and correct diagnosis). Requiring specialists to better

justify diagnoses, particularly in the presence of disconfirming evidence, has been found to decrease overconfidence in other domains.²³ Actively discussing diagnostic possibilities with patients (asking them which of several diagnoses they think is most likely) and using debiasing techniques (cognitive strategies to overcome one's biases in judgment and thinking)²⁴ and reflective practice (critical self-deliberation about one's own decision making)²⁵ may also be useful.

Fifth, all specialists involved in the care of complex patients should be receptive to, indeed seek out, external feedback about any missteps in diagnosis that come to light over time, including after discharge from specialists' care. Lack of follow-up often leads to assumptions that diagnoses (and consequent treatments) have been appropriate. Being notified by GPs of diagnoses that subsequent events have shown to be incorrect, and specialists sending progress letters to referring GPs (especially when re-referrals are being requested) inviting them to give their opinion on the appropriateness of current diagnoses may also improve diagnostic calibration. One study showed that half of patient-initiated second opinion requests to specialists related to diagnosis, and of these, the diagnosis was changed in 15% of cases overall, of which 20% had moderate to major clinical impact.²⁶ Certain specialties, such as gastroenterology, neurology and rheumatology, attracted higher than average rates of diagnosis change (22–26%) with higher rates of moderate and major impact (22–27%).²⁶

Finally, health care is moving from a disease-centred model towards a patient-centred model, and this will increasingly blur the boundaries and accountabilities between the different specialties in regards to diagnostic expertise applied to patients with multimorbidity. Greater interspecialty communication and more continuous oversight of the care of individual patients by GPs and specialists with generalist training will be required in minimising diagnostic error.

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