Diagnostic error: incidence, impacts, causes and preventive strategies

Some form of diagnostic error occurs in up to one in seven clinical encounters, and most are preventable

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Diagnosis consists of eliciting information from history and examination, formulating a differential diagnosis, and selecting a final diagnosis based on the predictive value of specific clinical features and laboratory investigations. A timely and accurate diagnosis is every patient's expectation.

Prevalence, impacts and causes of diagnostic error

Diagnostic error comprising missed, wrong or delayed diagnoses (Box 1) affects between 8% and 15% of all hospital admissions in the United States,[1,2] with similar rates among patients with common diseases attending outpatient clinics.[1] As many as 1.1% of adult hospital admissions will involve diagnostic error that causes harm to patients.[3] Nearly a third of all preventable deaths in acute hospitals in the United Kingdom are attributed to diagnostic error.[4] In Australia, an estimated 140 000 cases of diagnostic error occur each year, with 21 000 cases of serious harm and 2000–4000 deaths.[5] Almost one in two malpractice claims against general practitioners involves diagnostic error.[6]

More than 80% of diagnostic errors are deemed preventable.[7] Cognitive factors in clinician decision making are primary or contributory causes of more than 75% of diagnostic errors, with system errors (eg, missed communication or follow-up of a laboratory test result) being less frequent.[1] Failure to formulate an adequate differential diagnosis[8] and overconfidence in incorrect diagnoses[9] are major contributors. Clinical culture discourages disclosure of diagnostic errors and they are largely neglected within professional training curricula[10] and organisational quality and safety programs.[11]

Identifying the cognitive causes of diagnostic error which can inform preventive strategies requires an understanding of clinical reasoning (Box 2).[12-15] Intuitive thinking is the preferred reasoning mode, using heuristics (ie, mental shortcuts or rules of thumb) to accelerate the process by limiting the load on short term working memory to no more than seven ideas at a time. While efficient and accurate in many situations, heuristics can be misapplied due to cognitive bias (Box 3). Emotions, fatigue, distractions, peer opinions, and cultural norms can also further impair cognitive fidelity.

Strategies to prevent diagnostic error

Various preventive strategies have been proposed, the choice of which may vary according to clinician experience, types of clinical scenarios encountered, and the clinical environment.

Optimise the clinical interview

Taking a good history, including collateral information from relatives and other health professionals, and performing an adequate physical examination are fundamental. In combination, these will yield the correct diagnosis in more than 80% of cases,[16] while failure to enact them contributes to 40% of missed diagnoses.[5,17]

Target education to specific scenarios commonly associated with diagnostic error

Knowledge deficits are infrequent (< 5%) causes of diagnostic error among practising clinicians.[18] It is not that clinicians are unfamiliar with a diagnosis, they simply fail to consider it when appropriate. Educational interventions to increase overall knowledge do not necessarily improve diagnostic performance.[19] More useful is tuition focused on scenarios involving frequently missed or wrongly diagnosed conditions, including vascular events, infections, cancer, and neurological disorders (eg, multiple sclerosis).[20] Targeted training, such as how to recognise subarachnoid haemorrhage,[21] has prevented some condition-specific diagnostic errors.

Verify past diagnostic labels

Between 11% and 40% of listed diagnoses in older patients with Parkinson disease, dementia, heart failure and chronic obstructive pulmonary disease do not satisfy accepted diagnostic criteria.[22] Verification of past diagnoses, especially those based solely on subjective judgements and lacking specific diagnostic tests, is needed when clinical trajectories are atypical or appropriate therapies yield no response.

Implement strategies for reducing cognitive errors

Recent reviews describe various strategies for reducing cognitive errors[23-25] with varying levels of evidence of efficacy.

Lectures, seminars, group discussions, and interactive videos can all improve knowledge of cognitive biases and debiasing strategies, broaden differential diagnosis, and enhance reasoning processes. However, evidence of improved diagnostic accuracy is lacking, suggesting that, despite such educational interventions, clinicians may still not reliably identify when biases are influencing diagnostic decisions.

Diagnostic checklists can take various forms:

- a generic checklist prompting clinicians to optimise their cognitive approach;

1 Typology of diagnostic error

Diagnostic errors can be of three types:

- missed diagnosis — the correct diagnosis was never considered;
- wrong diagnosis — the provisional or working diagnosis is incorrect;
- delayed diagnosis — sufficient information was available to enable the correct diagnosis, which was eventually made, to be made at an earlier time.

The term "overdiagnosis" refers to a separate concept where a diagnosis is correct (eg, a patient has prostate cancer) but the diagnosed condition is not causing symptoms, is of low grade of malignancy, and will not prematurely kill the patient before they die of other diseases. In this scenario, the very act of diagnosing this disease may actually cause harm by invoking needless clinical intervention. It is different to when a diagnosis is actually incorrect, which is the focus of this article.

- a differential diagnosis checklist prompting clinicians to consider the correct diagnosis as a possibility; and
- a checklist of common pitfalls and cognitive forcing functions for evaluating particular diagnoses, including cross-checking with patients.[26]

Only the differential diagnosis checklists show improvements in the completeness of differential diagnosis in simulated or actual cases.[27] In one study, a differential diagnosis checklist led to fewer errors overall;[28] another similar tool combined with a debiasing checklist increased diagnostic accuracy compared with intuitive reasoning.[29]

Cognitive forcing strategies, defined loosely as any form of disciplined thinking, require clinicians to consciously slow their thinking and systematically evaluate all potential alternatives and mimics before finalising a diagnosis.[30] In some studies,[31] but not others,[32] this approach improves diagnostic accuracy compared with first impression diagnoses or reasoning without any specific instruction. In one study, instructing participants to reconsider their diagnosis after removing a distracting detail from the case outline greatly improved diagnostic accuracy.[33] Similar to cognitive forcing strategies, analytical reasoning involves instructing participants to use a guided, analytical approach (System 2) rather than rapid intuition (System 1). Diagnostic accuracy improves,[34,35] more so when dealing with complex cases,[36] and in a randomised trial,[35] this approach overcame deliberate attempts within test cases to induce cognitive biases.

Deliberate practice actively engages clinicians in solving diagnostic conundrums (real or vignette) and verbalising their reasoning ("thinking out loud") as the case unfolds.[37] By comparing participants' reasoning with those of an expert who has worked through the same case, cognitive errors and knowledge deficits can be identified. Simply seeing more cases, without any attempt at calibration, does not guarantee diagnostic expertise,[12] although whether deliberate practice improves diagnostic accuracy remains uncertain.

Metacognition involves clinicians thinking about their thinking and reflecting on past diagnoses and appropriate use of heuristics. In some studies, cued and modelled reflection improves diagnostic accuracy compared with a more generic, free-floating reflection[38] or leaving participants to reflect in whatever way they choose.[39]

Seeking second opinions on one's diagnoses from one's clinical peers can increase diagnostic accuracy by as much as a third.[40] Seeking the diagnostic opinion of patients, families and other members of the health care team, even if expressed in general terms, can also help detect and prevent errors.[41]

Following up patients over time, asking patients and colleagues to report errors, and implementing protocols for identifying errors (eg, trigger tools within electronic medical records for identifying unexpected adverse events or unplanned readmissions, or

2 Theories of diagnostic reasoning

Proponents of **organised (or structured) knowledge** emphasise content specificity whereby reasoning proficiency varies from case to case, depending on levels of knowledge of particular clinical scenarios. Clinicians construct multiple illness scripts as mental representations of diagnostic, therapeutic and prognostic attributes of specific conditions.[12] These scripts store and, with increasing experience, elaborate knowledge in a readily accessible format for application to new clinical scenarios. This emerging expertise is further developed by deliberate practice under supervision coupled with regular feedback.[13]

Proponents of **cognitive processing (or dual processing theory)** describe a rapid, intuitive form of pattern recognition (fast [System 1]) and a more deliberate, analytical approach (slow [System 2]).[14] When considering different or even single cases, clinicians oscillate between the two systems according to their level of experience and store of memorised patterns. Expert clinicians spend more time in System 1, novice clinicians more in System 2. Central to System 2 is the hypothetico-deductive model whereby the initial problem representation, gained from history and containing key clinical features (or cues), triggers a number of possible diagnostic hypotheses. These are ranked in decreasing likelihood and, based on further information from hypothesis-driven, focused physical examination and selected laboratory investigations, gradually eliminated in arriving at a provisional diagnosis.

The two schools of thought are not mutually exclusive and are in fact interdependent. Clearly, more hypotheses may be generated, or more patterns recognised, if the clinician can draw on a larger store of illness scripts that share cues with the problem at hand. Similarly, knowledge becomes more organised more quickly if clinicians consistently and systematically apply analytical thinking to obscure or atypical cases.

Approaches to improving diagnostic reasoning vary in their emphasis on expanding organised knowledge, mitigating cognitive bias, or optimising system of care factors according to how much each, in different circumstances, is considered the prime determinant of diagnostic error.[15]

3 Common cognitive biases in diagnostic reasoning

Bias	Definition	Example
Premature closure	Narrow rapid focus on single or a few clinical features in the clinical presentation to support a diagnostic hypothesis without considering other alternatives	Patient with rheumatoid arthritis who is receiving immunosuppressive medication presents with shortness of breath, inspiratory crackles on chest auscultation and diffuse fine infiltrates on chest x-ray. Congestive heart failure is quickly accepted as the diagnosis but subsequent bronchoscopy reveals *Pneumocystis* pneumonia
Anchoring bias	Tendency for clinicians to cling to their initial diagnostic hypotheses even as contradictory evidence accumulates	Patient with end-stage renal disease presents with altered mental status and myoclonus of the left arm, which is attributed to uraemia (the anchor). Failure of this syndrome to improve with dialysis (contradictory evidence) is underweighted until clinicians finally accept the eventual diagnosis of status epilepticus
Confirmation bias	Tendency to selectively search for features that support the initial or favoured diagnostic hypotheses rather than take deliberate note of features that challenge these hypotheses	Patient with past history of coeliac disease presents with symptomatic anaemia and low reticulocyte count, which is diagnosed as iron deficiency anaemia. Iron studies showing borderline low serum ferritin are interpreted as confirmatory evidence, while the finding of a widened mediastinum on chest x-ray is ignored. The patient is later diagnosed as having a thymoma associated with aplastic anaemia
Availability bias	Tendency to overestimate the probability of a diagnosis based on how easily it is recalled, which is often skewed by recent and memorable, or emotionally laden cases	A clinician who has recently seen a patient with myosarcoma who presented with left calf pain then begins to evaluate all subsequent similar presentations for the possibility of the same diagnosis
Representativeness bias/base rate neglect	Tendency to greatly overestimate the likelihood of a rare diagnosis on the basis of some prototypical features of that disease	Patient presenting with pulsatile headache, palpitations, diaphoresis and elevated blood pressure is diagnosed as having a pheochromocytoma (rare disease) whereas anxiety syndrome complicated by severe migraine (common disease) is later verified
Framing bias	Tendency for a presentation to be framed in a certain way according to past diagnostic labels (diagnostic momentum) or clinical setting (eg, medical *v* a surgical ward)	Patient with long-standing anorexia nervosa and post-traumatic stress disorder presents with weight loss, abdominal pain and diarrhoea. Her past history causes the clinician to frame the problem as one related to her mental health, leading to a diagnosis of irritable colon and laxative misuse associated with restrictive feeding. The presence of intermittent rectal bleeding and an elevated erythrocyte sedimentation rate (ESR) are underemphasised. The patient is eventually diagnosed as having Crohn's disease

systematic identification of errors within mortality and morbidity meetings) all provide information on final outcomes, thus checking the accuracy of initial diagnoses. Such strategies, combined with reflection on identified errors ("cognitive autopsies"), improve diagnostic performance.[42,43] Such feedback is important as clinicians' self-assessment of their diagnostic accuracy is unreliable and their level of diagnostic confidence can be insensitive to both accuracy and case difficulty.[9] Feedback also tempers over-reliance on the results of diagnostic tests that are at odds with the overall clinical picture and likelihood of a specific disease.[44]

High risk clinical environments, in which diagnostic error is more likely to occur, require clinicians to be more vigilant about their reasoning in such circumstances.[45] Rushed clinical handovers, heavy caseloads, distractions and interruptions, caring for critically ill or complex multimorbid patients, interactions with uncooperative or non-communicative patients, and clinician fatigue or personal stressors are some examples.[46]

Computer-assisted diagnosis in various forms can improve diagnostic performance. Computed decision support systems that generate differential diagnoses using inputted clinical data yield small improvements in diagnostic accuracy when clinicians revisit their diagnoses following a differential diagnosis generator consultation.[47] A digital image library of skin eruptions increased diagnostic accuracy of dermatology residents by 19% in a randomised trial.[48] An interactive computed decision support system achieved up to 75% reduction in diagnostic errors relating to vignettes of neurological disorders.[49] A web-based system that facilitated internet crowdsourcing of multiple opinions improved diagnostic accuracy among junior physicians.[50]

Acknowledging, explaining and sharing diagnostic uncertainty with patients helps to protect clinicians from rushing to ill-considered diagnoses. Up to 40% of first-contact primary care consultations involving a diagnostic question do not yield a definite answer.[51] In such situations, clinicians may feel pressured to prematurely commit to a diagnosis in order to activate management plans and demonstrate competence. In contrast, patients welcome an open discussion of possible differential diagnoses and a plan and timeline for ongoing review.[52] Injudicious ordering of multiple diagnostic tests to reduce uncertainty does not reduce patient anxiety and may cause harm from false positive results.[53]

Need for more research into diagnostic reasoning

While we have sought to shed light on the causes and prevention of diagnostic error, we concede current research has several limitations:

- enrolment of predominantly novice rather than experienced clinicians;
- non-randomised or before and after designs;
- relatively small samples;
- short term follow-up;
- variable methodological rigour;
- missing data; and
- multiple, often unvalidated, measures of error and reasoning style.

Primary outcome measures are restricted to improvements in knowledge or skills in vignette studies, although these are deemed reliable proxy measures of real-world decision making.[54]

Strengthening the evidence base for error mitigation is one objective of the recently established Australian and New Zealand Affiliate of the US Society to Improve Diagnosis in Medicine. This group aims to improve clinical diagnosis in this country with planned initiatives in practice improvement, research, education, and patient engagement (Supporting Information).

Conclusion

Despite limitations in current research, the scale and harm of diagnostic error obliges clinicians to consider adopting preventive strategies that have reasonable face validity, are easily implementable in workplaces, and target individual decision making.

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References are available online.

Supporting Information

Additional Supporting information is included with the online version of this article.

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