Overcoming Diagnostic Errors in Medical Practice

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The National Academies of Sciences-Engineering-Medicine report *Improving Diagnosis in Health Care* highlights the significant degree to which diagnostic errors impact clinical medicine. Although precise definitions of diagnostic error and comprehensive estimates of its incidence and impact are lacking, analysis of adult healthcare and postmortem examinations implicates diagnostic error in up to 17% of hospital adverse events and in approximately 10% of patient deaths.[1] There is a paucity of objective data on diagnostic error in pediatrics, although just more than one-half of surveyed pediatricians report making diagnostic errors at least once or twice monthly, and 45% of those surveyed describe diagnostic error leading to patient harm at least once or twice per year.[2] In intensive care settings, major diagnostic errors were found in up to 20% of autopsied pediatric and neonatal intensive care unit deaths.[3] Although quality and patient safety initiatives have highlighted the role of systems-related medical errors in patient outcomes, diagnostic error has remained a "blind spot" because such errors rarely are detected or reported.[1] Learning from diagnostic errors is even rarer.[4,5]

In many circumstances, diagnostic error is the result of human rather than systems errors.[6] Such errors in clinical reasoning are to occur most likely when individual clinicians invoke heuristics—pattern recognition and "rules of thumb"—oftentimes to the exclusion of more deliberate analytical approaches.[7] Although diagnostic errors are a risk in all patient encounters, the risk is greatest among patients with complex, multisystem, fragmented, or progressive symptoms.[8] In pediatrics, this challenge often is compounded by the wide variation in physiology and development of patients from birth through adolescence, in which both risk factors for disease and the actual manifestations of disease can vary by age.[9-14] In this paper, we will discuss our experience with patients who have had an inaccurate or delayed diagnosis who were referred to our Undiagnosed and Rare Disease (URD) Program. The evaluation of patients with undiagnosed and rare diseases requires consciously acknowledging potential biases or mistakes in clinical reasoning that can increase the risk of a cognitive diagnostic error. The experiential knowledge gained in our URD program has revealed a process and an environment that can mitigate these risks, improve outcomes for all patients, and offer insights into further study on the nature of and solutions to diagnostic errors.

A Definitional Framework of Medical Errors

Various conceptual frameworks for medical errors have been proposed. The National Academy of Medicine divides medical errors broadly into errors of execution, in which an appropriate plan fails to be completed properly; errors of commission, in which a planned action is ill-suited to achieve its aim; and errors of omission, in which a suitable plan is either not considered or not undertaken.[15] The Academy defines 4 specific subcategories of medical error: diagnostic, treatment, preventive, and other. Diagnostic error may consist of unintentional diagnostic delays, wrong diagnoses, or missed diagnoses.[16]

Diagnostic Error and Cognitive Diagnostic Error

Whether the diagnostic error consists of a delayed diagnosis, a wrong diagnosis, or a missed diagnosis, conceptual frameworks for the cause of diagnostic error include no-fault, systems-related, and cognitive errors.[16] No-fault errors include instances in which diseases present with masked or unusual manifestations, or in which diagnosis is obscured secondary to a patient being uncooperative with the evaluation. Systems-related errors include instances in which technical or organizational factors impede diagnosis, such as the unavailability of a particular diagnostic assay or the mislabeling or mishandling of a diagnostic specimen leading to erroneous results. Cognitive diagnostic errors are inherently related to clinicians and reflect faulty knowledge, data gathering, or synthesis.[16] Diagnostic errors rarely occur in isolation; an analysis of 100 cases of diagnostic errors found an average of 5.9 errors per case. Cognitive diagnostic errors were implicated most frequently: even though no-fault errors were the sole error type in 7% of cases, and 19% of cases were attributable to systems-related factors alone, 28% of cases were related to cognitive factors alone. Errors are more commonly multifactorial: the remaining 46% of cases were attributable to a combination of systems-related and cognitive factors, with 74% of all cases being related to either cognitive factors alone or a combination of systems-related and cognitive factors.[16] Given the significant contribution of cognitive errors to both diagnostic error and adverse patient outcomes in general, approaches to minimizing cognitive diagnostic errors should be based on an understanding of the etiology of these errors, which in turn is based on an understanding of clinical reasoning and medical decision-making.

Clinical Reasoning: Heuristic and Analytical Processes

Cognitive diagnostic errors reflect failures in medical decision-making, the conceptualization of which is based on dual process

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theory, which holds that clinicians engaging in medical decision-making invoke 2 distinct, yet occasionally simultaneous processes: a rapid, intuitive, and nonanalytical system 1 process frequently based in heuristics, as well as a more deliberate and analytical system 2 process that relies on hypothetical and counterfactual reasoning, in which potential explanations for clinical phenomena are explored in detail and are challenged with alternate explanations.[1] Given the time pressures and competing priorities clinicians often face, most rely on intuitive system 1 processes to a greater degree than analytical system 2 reasoning.[2,7,17] Although these intuitive approaches can and often do save time, producing satisfactory results with respect to routine diagnostic accuracy and patient outcomes, they are by nature more prone to fail when patient presentations are complex, multisystem, or evolving; when they fail, these heuristic processes become a form of bias that can lead to cognitive diagnostic error.[17]

Cognitive Biases in General Medical Practice

Although cognitive diagnostic errors may be the result of faulty knowledge or skills, such as a clinician misreading an electrocardiogram, knowledge or skills-based errors are implicated less frequently than are errors related to cognitive biases.[16] Cognitive biases are heterogeneous, may be introduced at any point in the diagnostic evaluation, and typically take 1 of 3 forms: heuristic failure, in which intuitive system 1 processes fail; errors of attribution, in which subjective internal factors related to personality or cultural characteristics influence medical decision-making; and errors of context, in which subjective external factors related to the setting of the diagnostic evaluation influence both intuitive and analytical approaches.

Cognitive Bias Related to Heuristic Failure. Diagnosis based on system 1 processes uses heuristics, in which particular associations among historical features, symptoms, and physical findings cue illness scripts, or preformed cognitive representations of specific diseases. Under ideal circumstances, information gathering in the initial stages of patient evaluation is robust enough to generate several illness scripts simultaneously, and the clinician can rapidly compare the unique features of the patient against these competing scripts to arrive at a sufficiently narrow differential diagnosis or at a specific diagnosis. In practical terms, however, the successful use of illness scripts depends highly on a clinician's previous experiences with the diseases in question, the ability to elicit a complete-yet-concise history and physical examination, and proper recollection and recognition of the nuances of both the disease processes being considered and the patient's individual circumstances.[18] Furthermore, heuristic processes are particularly prone to error when patient presentations are atypical.[7] Examples of this type of heuristic failure leading to diagnostic error may include a child with appendicitis whose abdominal pain fails to migrate to the McBurney point and who is diagnosed erroneously with constipation, or may be as complex as an infant with Kawasaki disease presenting with only fever.[19] When heuristic processes fail, they constitute a form of cognitive bias and can lead to diagnostic error.[1] Common forms of cognitive bias and diagnostic error related to heuristic failure are listed in **Table I**.[1,20-23]

Cognitive Bias Related to Errors of Attribution. Personality or cultural factors, both with respect to the culture of medicine and societal culture at large, may introduce cognitive bias into both system 1 and system 2 thought processes. The fundamental cognitive flaw under these circumstances is one of attribution, in which the clinician places undue importance on the perceived internal characteristics or motivations of others, whether they are the patient, the patient's family, or other members of the evaluation team. Primary among these subjective influences within the culture of medicine is the appeal to authority, in which the opinion of senior or otherwise expert clinicians is granted greater weight in the evaluation, regardless of whether that opinion is evidence-based or aligns with the objective findings of the patient. Authoritative statements from senior clinicians such as "I have never seen this symptom in the proposed disease" or "it must be this disease" can lead to a diagnostic momentum that can exclude consideration of other disorders. Influenced by these statements, other members of the evaluation team may engage in anchoring, in which the initial presenting features that informed the

Table I. Cognitive biases related to heuristic failure

Biases	Definition
Anchoring	Locking into a diagnosis based on initial presenting features, failing to adjust diagnostic impressions when new information becomes available.
Confirmation bias	Looking for and accepting only evidence that confirms a diagnostic impression, rejecting or not seeking contradictory evidence.
Diagnostic momentum	Perpetuating a diagnostic label over time, usually by multiple providers both within and across healthcare systems, despite the label being incomplete or inaccurate.
Expertise bias/yin-yang out	Believing that a patient who has already undergone an extensive evaluation will have nothing more to gain from further investigations, despite the possibility that the disease process or diagnostic techniques may have evolved so as to allow for appropriate diagnosis.
Overconfidence bias	Believing one knows more than one does, acting on incomplete information or hunches, and prioritizing opinion or authority, as opposed to evidence.
Premature closure	Accepting the first plausible diagnosis before obtaining confirmatory evidence or considering all available evidence. "When the diagnosis is made, thinking stops."
Unpacking principle	Failing to explore primary evidence or data in its entirety and subsequently failing to uncover important facts or findings, such as accepting a biopsy report or imaging study report without reviewing the actual specimen or image. Especially important in undiagnosed and rare diseases.

Table extracted from Kliegman et al,[20] Committee on Diagnostic Error in Health Care, Board on Health Care Services, Institute of Medicine, The National Academies of Sciences, Engineering, and Medicine,[1] Croskerry,[21,22] and Rencic et al.[23]