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Imperatives, expediency, and the new diagnosis

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Diagnostic principles are no different today than they were in the past, perhaps even for Hippocrates. We still generate diagnostic hypotheses early in the course of a patient encounter, often with precious little information. We are still guided by these early hypotheses to gather data that help us to iterate toward an accurate diagnosis. We still create rolling differential diagnoses, and modify them repeatedly as we apply diagnostic tests in accordance with our sense of disease likelihoods. And we assemble working diagnoses based on our confidence that we have explained all the abnormal findings, pathophysiologic correlations, and causal connections [1, 2]. But the comparison between the diagnostic process of yesteryear, especially as it applies to hospitalized patients, and that of today, ends there. Today's diagnosis bears little resemblance to how the process proceeded even as recently as one or two decades ago. The major factors responsible for this rapid evolution in diagnostic strategy are the quality and cost of medical care.

To begin with, the character of diagnostic tests and treatments has changed enormously. Focusing on imaging illustrates the issue. Scans, including ultrasound, CT, MRI, and their derivatives now identify internal structures and lesions that were hitherto invisible and inaccessible. Though they are expensive, are nearly always applied far more often than originally intended, and frequently uncover puzzling false positive findings that require unnecessary further investigation, there is little doubt that scanning spares patients from interventions that, in the past, yielded discomfort and risk. Moreover, they often quickly provide an "answer," thus short-cutting a long and tedious workup and expensive hospital days. It has become commonplace, especially in emergency departments, to take a brief history, check vital signs, and immediately send the patient for a scan in the expectation that the diagnosis will become evident even before a complete workup is carried out [3]. Scanning has also dramatically changed patients' and physicians' attitudes toward tissue biopsies. Whereas the risk of bleeding, organ perforation, and need for repeated punctures to obtain adequate specimens were often deterrents to obtaining material from organs, fluid collections, and deep lesions, there are few locations in the body inaccessible now, and the risks of adverse events following the procedures have become more acceptable, at least measured against the benefit of the information obtained [4].

The simultaneous evolution of therapy has been no less revolutionary. Whereas patients with acute myocardial infarctions, for example, were previously kept at bed rest for weeks with the hope that their hearts would heal, now they are quickly identified in the field, rushed to the nearest hospital, and whisked off to the catheterization laboratory with the result that myocardium at risk is converted to viable tissue – thus avoiding months or years of heart failure-related dyspnea [5]. Similarly, acute stroke is no longer an indication for watchful waiting but an imperative for swift intervention [6]. Many other clinical conditions that benefit from rapid, accurate diagnosis come to mind, including vasculitis with acute renal failure, infectious endocarditis, pulmonary emboli [7], and thrombotic thrombocytopenic purpura [8]. Disorders such as these and the success of immediate intervention are the imperatives that shape the diagnostic strategies of today.

The practical implementation of rapid diagnostic workups has implications in turn for how certain tests, especially scans are used, where they are ordered, and how much we spend on them. Because of the slow and silent decline of test risk and advances in therapeutics, little attention has been paid to their consequences, both in terms of the quality and cost of care. What, for example, is the tradeoff in the quality of care and the expense of the total hospital stay between quickly scanning patients presenting to a hospital for various acute conditions versus following the traditional practice of the systematic workup based on serial assessment of information obtained from the patient's history, physical examination, and performance of routine blood and urine tests? A quickly performed scan might be expensive, but it also might provide a working diagnosis immediately, shorten an otherwise longer hospital stay, and conserve the hospital's resources [9]. (Then, again, an unexplained finding might entail further workup and risks that would never have been experienced if the scan had not been done in the first place.) If we knew which conditions and which

tests were likely to lower risk and improve efficiency of workups, we might be able to satisfy the two modern hallmarks of health care innovation, namely improving the quality of care and simultaneously lowering cost [10]. To do so, however, we will have to discover who it is that makes the testing decisions and the soundness of their choices. We will need solid data on efficiency, risk, and cost of tests and treatments to answer these questions. We will also have to reconsider when in the sequence of a diagnostic evaluation invasive studies are most appropriate. If it is apparent at the time of admission, for example, that the performance of a kidney or liver biopsy or ERCP is inevitable, why not carry out the procedure immediately instead of awaiting other test results?

As with all innovations, we must be prepared for unintended consequences. Would earlier scanning become a mindless, knee-jerk process devoid of cognitive input? What are the implications for teaching medical students and residents the appropriate indications for use of diagnostic tests? What are the implications for the teaching of clinical reasoning, including the appropriate use of drugs and devices? How can we preserve the intellectual satisfaction in the challenges of the diagnostic process if we advance further into territory already occupied by thousands of practice guidelines? What are the implications of attracting students and residents into the so-called cognitive specialties? Researchers need to begin to explore these issues.

It is clear, however, that diagnostic testing can no longer be considered in terms of the cost of the procedure, but in the overall context of the total cost of a hospital stay or a bundled payment. Moreover, we must pay more attention to demarcating those diagnostic categories that, because of their therapeutic imperatives, should receive priority when it comes to imaging requests. Although satisfying the expediency of hospital operations could further rigidify and mechanize the role of physicians, diagnosis will always be an intellectually challenging task because most diseases do not come in consistent packages, and patients differ in how they respond. Diagnosis remains fundamentally dependent on a personal interaction of a doctor with a patient, the sufficiency of communication between them, the accuracy of the patient's history and physical examination, and the cognitive energy necessary to synthesize a vast array of information.

We need not give up probabilities, likelihood ratios, expected utility, or Bayes' Rule, but we must evolve our concepts of diagnosis in light of today's clinical exigencies, therapeutic imperatives, and cost.

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