REVIEW

THE AMERICAN JOURNAL *of* MEDICINE®

How Expert Clinicians Intuitively Recognize a Medical Diagnosis

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ABSTRACT

Research has shown that expert clinicians make a medical diagnosis through a process of hypothesis generation and verification. Experts begin the diagnostic process by generating a list of diagnostic hypotheses using intuitive, nonanalytic reasoning. Analytic reasoning then allows the clinician to test and verify or reject each hypothesis, leading to a diagnostic conclusion. In this article, we focus on the initial step of hypothesis generation and review how expert clinicians use experiential knowledge to intuitively recognize a medical diagnosis.

 • *The American Journal of Medicine (2017) 130, 629-634*

KEYWORDS: Exemplars; Experiential knowledge; Heuristics; Illness script; Intuition

A medical diagnosis, such as acute myocardial infarction, often is defined by rules or criteria that are codified by panels of experts.[1,2] At the outset of making a diagnosis, however, those rules could not be further from the mind of a clinician. Rules are used for analyzing and verifying a diagnosis, but the initial diagnostic step used by expert clinicians is not rule based. The ability of expert clinicians to recognize a possible diagnosis is intuitive, nonanalytic reasoning.[3,4]

More than 4 decades ago, a number of researchers independently deconstructed the diagnostic process and found that expert clinicians use the same process of hypothesis generation and verification that is generally used for reasoning by beginning medical students.[5-8] The skill of master diagnosticians was not due to a distinctive reasoning process, but instead depended on a clinician's ability to access knowledge from past experience to generate short lists of possible diagnoses. Elstein et al[5] and Barrows et al[7] noted that expert clinicians developed 3 to 5 hypotheses within seconds to minutes of starting a diagnostic inquiry. Barrows et al[7] showed that early hypothesis generation was critical to the accuracy of the eventual diagnosis. If the clinician thought of the correct diagnosis within 5 minutes, eventual accuracy was 98%; if not, accuracy decreased to 25%.[7] In a subsequent study of primary care physicians, the correct diagnosis occurred to the clinician on the basis of only the chief symptom in 78% of cases.[9] A study of emergency physicians showed that clinicians generated 25% of the diagnostic hypotheses before even meeting the patient and 75% of the hypotheses within the first 5 minutes of the clinical encounter.[10] The cognitive psychologist Herbert Simon described this astonishing human ability by stating: "the situation has provided a cue; this cue has given the expert access to information stored in memory, and the information provides the answer. Intuition is nothing more and nothing less than recognition."[11]

DIAGNOSTIC CATEGORIES

Generating diagnostic hypotheses is a categorization process. Psychologists describe how various decision-makers form categories and how they place new objects into those categories. According to psychologist Douglas Medin, "a category is a partitioning or class to which some assertion or set of assertions might apply."[12] A diagnostic category is useful because it allows the clinician to make inferences and predictions about patients assigned to the diagnostic category.

Funding: None.

Conflict of Interest: None.

Authorship: All authors had access to the data and played a role in writing this manuscript.

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0002-9343/$ -see front matter
http://dx.doi.org/10.1016/j.amjmed.2017.01.045

When placing a patient with specific signs and symptoms into a category (eg, myocardial infarction), experts with prior experience can immediately recognize that acute myocardial infarction is a diagnostic possibility because the patient resembles a prior patient with acute myocardial infarction. This ability to place a patient in a diagnostic category is similar to the general ability to place a common object such as a bird, dog, or chair into a category. People can recognize a variety of birds, ranging from a robin to a penguin, because they have seen a variety of birds before and have placed them in memory under the category of birds. Likewise, expert clinicians can recognize a broad range of patients with acute myocardial infarction because they have seen a variety of patients with acute myocardial infarction before and have placed them in long-term memory under the category of acute myocardial infarction.

Diagnostic possibilities are not always immediately recognizable, requiring the clinician to connect the clinical cues, like piecing together a jigsaw puzzle. To help this process, clinicians are taught to take a patient's history and organize it into a narrative.[13-16] A narrative is a way of organizing a story by adding context and detail, and assigning priority and weight to elements of the narrative. When an expert clinician retells the story, the patient's words become the doctor's words. "I'm short of breath" becomes "dyspnea," and "I broke out in a sweat" becomes "diaphoresis." The experienced clinician will add semantic qualifiers, which are meaningful adjectives such as "acute" or "chronic," and "episodic" or "continuous" to fully describe a clinical presentation.[17] Humans are natural storytellers, and often the diagnosis is more recognizable when the patient's story is organized as a coherent narrative.

Abstracting the meaning from the patient's history and re-representing the patient's findings as a succinct summary or problem statement is another opportunity to recognize that the patient belongs to a diagnostic category. In ill-defined, complex cases, pattern recognition may not be sufficient, and experts will adapt by relying more on analytic reasoning based on causal or conceptual knowledge. Clinicians may consult other specialists, bringing additional experiential and conceptual knowledge to bear to solve difficult cases.[18,19]

CLINICAL SIGNIFICANCE

- Expert clinicians use intuitive, nonanalytic reasoning to generate approximately 3 to 5 diagnostic hypotheses early during a diagnostic encounter.
- Prior experience, structured in long-term memory as exemplars, enables clinicians to automatically and effortlessly recognize diagnostic hypotheses.
- Research is pointing to ways that learners can make the most of experience to optimize the way they use experiential knowledge to generate diagnostic possibilities.
- Research provides guidance for educators and clinicians on what works and what does not work for improving the diagnostic process.

HOW KNOWLEDGE OF PAST EXPERIENCE IS STRUCTURED

If the ability to recognize a diagnosis is less dependent on a distinctive reasoning process and more dependent on knowledge of past experience, it is important to understand how memories of past experiences are stored and retrieved. Investigators have promoted a number of theories to explain how we structure experiential knowledge.[20-27] Knowledge is remembered in context, and experiential knowledge gained from direct experience is structured differently than formalized biomedical knowledge gained from abstract (eg, book) learning. Experiential knowledge is remembered as instances of past experience, often referred to in the psychology literature as "exemplars": prior experiences that have been categorized and stored in memory.[22] Assigning an experience to a category gives it meaning, a process called "instantiation." With experience, a category will contain a number of exemplars that are stored in a nearly limitless long-term memory and are automatically retrievable.

Each clinical encounter is represented in long-term memory in a unique and idiosyncratic fashion, attending to key features of the presentation that hold meaning specific to that clinician. This representation may contain both the relevant features of the disease and other features unique to this specific person. Because each clinician has unique patient experiences, the exemplars available to each clinician is a product of his or her unique experiences and not generalizable among clinicians.

When encountering a patient with chest pain, an expert may recognize that the patient is a variant within the category of acute myocardial infarction (eg, posterior myocardial infarction or non−ST elevation myocardial infarction) or is a variant of myocardial infarction mimics (eg, type A aortic dissection or pericarditis) based on pattern recognition and the degree of association with existing exemplars in each category. As learners become experts, exemplars can be compared and contrasted to help the learner remember how different disease categories have overlapping and distinguishing features.[28]

Patient presentations vary. Some patients have many disease features, and some have only a few. Exemplars help us handle the variation of disease presentation and allow us to recognize patients who lack all of the typical features of a disease.

Common presentations are repeatedly encountered, resulting in more numerous exemplars that are recalled more readily. This gives the expert an intuitive sense of the base rates of features within a category.[24] Experienced clinicians know intuitively that patients with acute myocardial infarction frequently present with chest pressure radiating to the arms, and among patients presenting with chest pain, acute myocardial infarction is more common than aortic dissection.

Complementary to exemplars, psychologists have proposed the theory of illness scripts.[29] An illness script is a schema or map, unique to the clinician, integrating and linking various nodes of information to describe a general presentation. For example, an illness script for an undifferentiated chest pain presentation would link previous clinical experiences (ie, exemplars) with formalized knowledge (eg, pathophysiology, epidemiology). Navigating the script allows the clinician to identify features of the presentation that are consistent or inconsistent with a diagnosis such as acute myocardial infarction or aortic dissection.

Novice medical students lack experience and rely on causal reasoning to generate diagnostic hypotheses. As trainees gain experience, they begin to use illness scripts. As clinicians become experts, they become more reliant on exemplars for early hypothesis generation.[29,30]

Regardless of whether early hypothesis generation is best explained via exemplars, prototypes, or illness scripts, it is clear that expertise requires authentic clinical experiences informed by feedback on patient outcomes.[31] The association of a new case with a past experience improves the richness of the diagnostic category and increases the accuracy and speed of future recall of this information in making a new diagnosis.

DUAL PROCESS THEORY

Dual process theory is one explanation of how we use a combination of intuition and analytic reasoning to solve problems in general and diagnose patients in particular.[4,32,33] According to this theory, we use 2 definable systems of thinking called "System 1" and "System 2." System 1 thinking is intuitive, automatic, quick, and effortless. System 2 thinking is analytic, reflective, slow, and effortful. For making a diagnosis, System 1 enables the quick retrieval of an exemplar stored in long-term memory to bring to mind a diagnostic possibility. This occurs automatically, naturally, and without conscious control. System 2 is used for testing, analyzing, and verifying a diagnostic hypothesis. This requires focus, attention, and cognitive effort. Functional magnetic resonance imaging studies have shown that System 1 and System 2 thinking involve different areas of the brain and have different metabolic requirements.[34,35]

System 1 thinking frequently makes use of heuristics. This term, derived from the Greek word *heuriskein* (to discover), is defined as an aid to problem solving that is learned through experience. Cognitive psychologists use the term to describe a mental shortcut that enables rapid decision-making under conditions of uncertainty.[4,11,36]

Heuristics were promoted as a tool for solving problems in a book entitled *How to Solve It*, written years ago by George Polya,[37] a Stanford mathematics professor. Polya taught students the habit of using a variety of simple mental processes to approach mathematics problems. He encouraged students to ask questions such as, "What is the nature of the problem? What is known and what is unknown? Have I seen anything like this before?" By developing the habit of using these short mental processes, the processes became intuitive, developing into useful heuristics.

Heuristics were the topic of investigation by Herbert Simon,[11] working in the field of artificial intelligence. Simon described 2 general types of problems: unstructured problems and structured choices. According to Simon, heuristics are used for unstructured problems to organize cues, narrow the problem space, and simplify a seemingly intractable problem into a manageable series of structured choices. Simon's general concept of heuristics is useful for understanding diagnostic reasoning because patients often present with problems that are initially unstructured and undifferentiated. Heuristic-based, nonanalytic reasoning enables the expert clinician to initially sift and sort through diagnostic information. Early hypothesis generation enables the clinician to narrow the search and become more focused on the problem at hand. Once the hypothetical diagnostic possibilities are identified, the problem becomes a structured choice solved by further evaluation and diagnostic testing using System 2 analytic reasoning.

In medicine, physicians are trained to ask, "What could this problem be? Is it localized or systemic? What else could it be? What critical diagnosis can I not miss?"[13-16] These questions seem to bridge System 1 and System 2 thinking, in a manner similar to Polya's notion of heuristic problem-solving.[37]

Heuristics often are linked to cognitive biases. Kahneman[4] and Tversky, among others,[36] proposed that heuristics represent a speed and accuracy tradeoff, in which their speed may lead to bias and error. Lopes[38] has argued that most of the research on heuristics and biases used artificial experimental methods that do not represent the environment in which decision-makers actually perform. Many of the experiments informing the literature on bias were performed on convenience samples of undergraduate psychology students and not on experts, neglecting the role of expertise in heuristic-facilitated decision-making.[39] A full discussion of heuristics and biases is beyond the scope of this review, but the availability heuristic bears mentioning. This heuristic describes how decision makers judge events that are more readily remembered as being more frequent. This heuristic is useful for clinicians in practice, but Kahneman[4] and Tversky have described how a recent or salient memory can sometimes bias a decision maker's frequency estimate. Most clinicians can remember an episode when a particularly memorable case (eg, a missed aortic dissection) may have subsequently affected their judgment. The incidence of this bias in practice likely is not measurable, seems to be low, and was not observed in an experimental study.[24]

Several authors have implicated nonanalytic System 1 reasoning as the major source of diagnostic error.[40-42] However, research supporting this position is sparse. A recent review identified 213 studies of cognitive bias in both diagnostic and therapeutic decision-making. Only 15 of the studies addressed diagnostic decision-making, and only 7 biases were identified and examined.[43] Furthermore, studies have shown that cognitive biases tend to disappear as people develop expertise.[24,44]

Two studies were retrospective reviews of diagnostic errors in actual practice. These studies revealed mixed results about the role of System 1 and System 2 thinking. Graber et al[45] studied 100 cases of diagnostic errors in the emergency department and found that 68% of cases were associated with cognitive bias, primarily premature closure (ie, terminating the search for diagnostic hypotheses before collecting all of the necessary critical information).[45] However, premature closure would seem to be a breakdown of a System 2 process, rather than a System 1 error.[46] Another retrospective review of diagnostic errors in actual practice found diagnostic errors were more associated with knowledge deficits rather than with cognitive biases.[47]

Retrospective reviews can, themselves, be subject to hindsight bias and inter-reviewer variability. Indeed, a recent study showed that reviewers who claimed expertise in identifying biases showed poor agreement in identifying which bias was present.[48] In this study, investigators asked reviewers to evaluate cases in which half of the case vignettes implied an incorrect diagnosis and half implied a correct diagnosis. The reviewers identified twice as many biases for the case vignettes that seemed to lead to an incorrect diagnosis, indicating that these reviewers who were reviewing the cases to detect bias were themselves affected by hindsight bias.

IMPROVING DIAGNOSTIC ACCURACY

A recent Institute of Medicine report brought attention to the high prevalence of diagnostic error.[49] This report called for developing and deploying approaches to prevent diagnostic errors in practice and by enhancing education and training in the diagnostic process. The quality expert W. Edwards Deming[50] said, "If you can't describe what you're doing as a process, you don't know what you're doing." How can we improve the intuitive recognition of diagnostic possibilities?

Kahneman provides the following advice: "The way to block errors that originate in System 1 is simple in principle: recognize the signs that you are in a cognitive minefield, slow down, and ask for reinforcement from System 2."[4] Evans counters by stating, "perhaps the most persistent fallacy in the perception of dual-process theories is the idea that Type 1 processes (intuitive, heuristic) are responsible for all bad thinking and that Type 2 processes (reflective, analytic) necessarily lead to correct responses…So ingrained is this good-bad thinking idea that some dual-process theories have built it into their core terminology."[32]

Will slowing down prevent System 1 errors and improve diagnostic performance? A number of studies show that faster response times are more often associated with a correct diagnosis, suggesting that the additional time associated with deliberation (System 2) may not reduce errors.[51-54] When test subjects are instructed to go slow and be more analytic (deliberately overriding System 1 thinking with System 2 thinking), there is no effect on the accuracy of diagnosis.[51] Thus, Kahneman's[4] instructions to slow down and tame System 1 with analytic System 2 thinking may not apply to experts in medicine.

Some investigators have advocated educating physicians about potential biases and encouraging them to routinely de-bias their thinking in practice.[40-42,55] Research supporting this recommendation is sparse.[39] Three studies examining the effects of educational interventions to teach cognitive biases to medical trainees showed that the intervention had no effect.[56-58] There are no studies showing that active de-biasing is effective in practice. Given the long list of biases, the difficulty of clinicians to agree on the definitions of biases, and the difficulty in seeing one's own (unconscious) biases, a strategy of active de-biasing seems unlikely to improve diagnostic accuracy.

The ability to access experiential knowledge to inform a diagnostic hypothesis depends on both storage and retrieval of illness scripts and exemplars. Schmidt and Mamede's review[59] of the literature suggests that knowledge-oriented approaches to help learners connect presenting signs and symptoms with pathophysiologic mechanisms (through self-explanation or deliberate reflection) are promising. Comparing and contrasting cases to identify distinctions between look-alike disease presentations also seems useful. However, of the 48 articles reviewed, only 24 had empirical evidence and the interventions that were examined had small effects.

Finally, to improve the retrieval of past experience, investigators have proposed using checklists.[60] One study showed that a differential diagnosis checklist improved the diagnostic accuracy of medical students, whereas a de-biasing checklist had no effect.[61] Whether clinicians could use a checklist effectively in practice remains an open question.

It seems that there is no substitute for experience and knowledge for improving diagnostic accuracy. Retrieving exemplars to inform a diagnostic hypothesis may be effortless, but it takes sustained effort to develop these exemplars. Making the most of clinical experience (eg, broad exposure with feedback on patient outcomes) seems to be the key to improving the quality of making a diagnosis.

CONCLUSIONS

Cognitive science research over the past 4 decades has described the diagnostic process and how we use nonanalytic reasoning to generate diagnostic hypotheses. This research has emphasized the critical role of experiential knowledge, stored in long-term memory as a rich and individualized collection of exemplars of various diagnostic categories. The ability to rapidly access experiential knowledge is a hallmark of expertise. Knowledge-oriented interventions such as self-explanation, deliberate reflection, and checklists may improve diagnostic accuracy, but there is no substitute for experience gained through broad clinical exposure and regular feedback on patient outcomes. Improving diagnostic decision-making deserves greater emphasis in medical education and clinical practice. Further research is required to determine which strategies work and which do not work for improving the quality of diagnostic reasoning.

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