

Medical Decision Making

Harold C. Sox
Michael C. Higgins
Douglas K. Owens

**SECOND
EDITION**

 **WILEY-BLACKWELL**

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Second Edition

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To Jean and Kathleen
To Sara, Rachel, and Christopher
To Carol, Colin, and Lara
Their support has meant everything.

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Foreword

The maturation of medical science during the last half of the twentieth century was most impressive. Clinical trials displaced observational studies that typically consisted of a dozen or fewer patients; the pathophysiology and genetics of many diseases were discovered; and diagnostic and therapeutic methods advanced. Crude diagnostic tests such as cholecystography and barium enemas and risky tests such as air encephalograms, needle biopsies, and exploratory laparotomies were made obsolete by technology. Flexible tubes, some outfitted with lights and cameras, CT, MRI, and PET scanners, and sophisticated immuno-analyses of blood and other body fluids gained immediate acceptance. Many therapies that were formulated by grinding up organs, desiccating them, and hoping that they would correct a deficit were replaced by new, potent chemicals.

Clinical reasoning, the processes behind both diagnosis and medical decision making, including the complex tradeoffs between the risks and benefits of tests and treatments, lagged behind advances in medical science. In the run-up to the last quarter of the century, students learned how to reason about patient problems by observing expert clinicians at work, and (if they dared) by asking them why they ordered this test or that, why they gave one drug or another. Because this apprenticeship approach was not codified, objectified, or quantified, medical texts struggled to explain clinical reasoning, and students struggled to learn it. And when the evidence of confusion about the use of tests and treatments first emerged, alarm bells clanged. Researchers had discovered extreme variations in the use of tests and treatments from one community to another and in regions across the country without a corresponding benefit for patients. Irrational testing and treating had begun to contribute substantially to an impossible escalation in the cost of care.

During the last three decades of the twentieth century, clinician–scientists began to examine the processes of diagnosis and decision making with tools from other disciplines, including cognitive science, decision science, probability, and utility theory. From these diverse sources the clinical science of medical decision making was hatched. Elements of the diagnostic process were identified and a language for explaining and teaching diagnosis was formulated. Cognitive errors in diagnosis were sought and methods developed to avoid them. The critical importance of a probabilistic representation of diagnosis, in terms of prior probabilities, conditional probabilities, and likelihood ratios, was recognized and put to use in the form of a centuries-old formulation of Bayes' Rule. Decision analysis, a discipline formerly used by the military, was applied first to individual clinical problems, later to classes of

problems, and eventually to issues of cost and efficacy of tests and treatments. Before the end of the twentieth century, a science of medical decision making and a language for teaching it had been born.

Implementing the new science, however, proved more difficult than developing it. Skeptics averred that physicians' estimates of probabilities were often flawed, that applying Bayes' Rule was not easy, and that decision trees were either too simple (and thus did not represent a clinical problem sufficiently) or too complex (and thus could not be understood). Many wondered whether medicine could be convinced to adopt these new approaches and whether the average physician could be expected to use them in their day-to-day practices.

As the field has evolved, some of these questions have been answered. Now, in the second decade of the twenty-first century, Bayes' Rule is used to design clinical trials, to develop decision rules that help physicians judge whether to admit patients suspected of having an acute myocardial infarction, and to develop compiled strategies for diagnosing and treating pulmonary emboli, to name a few applications. Decision analysis has been used to formulate answers for individual patients' dilemmas, but this use is time consuming, expensive, and requires special expertise. Nonetheless, decision analysis has found extensive application in clinical practice guideline development, cost-effective analyses, and comparative effectiveness studies. A cadre of physicians has become sufficiently skilled in the methods to apply them in active clinical teaching environments and to integrate them into medical student and residency curricula.

It is legitimate to ask why a student or resident should spend the intellectual capital to learn these methods. The answer is compelling. First, they help in learning and teaching the process of diagnosis. Second, the principles of screening and diagnostic and management decision making become transparent from an understanding of Bayes' Rule, diagnostic and therapeutic thresholds, decisional toss-ups, and decision analysis. Subjecting such issues to rational examination improves decision making and, consequently, patient care. Moreover, because these methods are the basis for so many analyses of health practices, appreciation of their limitations provides a healthy skepticism of their applications. Lastly, the approaches are powerful tools to pass on the concepts to others, as well as critical templates to understand honest differences of opinion on controversial medical practices.

For the past 25 years, *Medical Decision Making* has been an ideal venue for developing a rich comprehension of these methods and for understanding how to approach diagnosis; the new second edition is even better. Its chapter on Bayes' Rule, for example, is exemplary, explaining the method in multiple different formats. The chapters on selection of diagnostic tests and decision analysis are meticulously crafted so as to leave little uncertainty about the methods. A new chapter on modeling methods is richly illustrated by actual analyses; the chapters on expected value decision making, utility assessment, and Markov modeling have been extensively revised.

In short, this book has been a standard of the field, and the new edition will continue its dominance. There is little doubt that in the future many clinical analyses will be based on the methods described in *Medical Decision Making*, and the book provides a basis for a critical appraisal of such policies. Teachers of medical decision making will require it; medical students will dig into it repeatedly as they learn clinical medicine; residents will go back to it again and again to refresh their diagnostic and therapeutic skills. And from its lucid pages, practicing physicians will attain a richer understanding of the principles underlying their work.

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Preface

The first edition of *Medical Decision Making* was a small project that took on a life of its own. The chapters began as sketches for a course on medical decision making that the authors undertook as part of a foundation grant to study methods for teaching medical decision making. Thanks to the enthusiasm of co-authors Keith Marton and Michael Higgins, the project took off and turned into a book published by Butterworths in 1988. A Stanford medical student, Marshal Blatt, read the chapters and gave us invaluable advice about making the book more understandable to beginning students. We must have listened to him because many students have thanked us for writing a book that they could understand.

Twenty-five years have elapsed since publication. *Medical Decision Making* has sold steadily through a succession of publishers. Physicians and decision analysts from every corner of the globe have approached me to say that the book was pivotal in their engagement with the field of medical decision making. People have called for a second edition, but the authors, having moved on in disparate careers, were never ready until the past year. I have been an advocate for medical decision analysis as a teacher, practicing internist, medical journal editor, and participant in the emergence of comparative effectiveness research as a new discipline. Michael Higgins worked for companies that developed medical software, while teaching courses at Stanford University. Douglas Owens, an internist and a leader in the application of medical decision analysis to clinical policy, has become the third author.

How has the book changed? Hopefully, the writing has benefited from my experience of eight years as a full-time medical journal editor. I updated Chapters 1 through 5, 9, and 11 and served as the editor for my co-authors. I rewrote my chapters, updated the examples, and added new developments (particularly a stronger emphasis on likelihood ratios (Chapter 4), systematic reviews and meta-analysis of diagnostic test sensitivity and specificity (Chapter 5), and cost-benefit analysis (Chapter 11)).

Michael Higgins wrote new chapters (Chapters 6 through 8) that covered expected value decision making, utility assessment, Markov models, and mathematical models of life expectancy. The treatment of these topics reflects his long teaching experience, in which he relied on mathematical models that simplify the process of assessing utilities. Any reader who can recall the concepts of high school algebra will be able to understand these chapters.

Dr. Owens' chapter on decision modeling reflects the growing influence of decision analysis to support clinical and public health policy making (and the waning influence of decision models created to solve a specific

patient's decision problem). The chapter describes different types of models and provides extended examples, but it is not a tutorial in how to create a decision model. The reader who wants to learn decision modeling should take the short courses offered at the annual meeting of the Society for Medical Decision Making and spend some time apprenticing with an expert in the field.

From the simple (likelihood ratios) to the complex (microsimulation modeling), what is the future of medical decision analysis? In two words, both hand-held computer applications ("apps" in current usage) and shared decision making. Hand-held devices will bring decision models – simple and complex – to the office and the bedside, where clinicians and patients will use them to individualize their discussions of the big decisions.

What can textbooks like *Medical Decision Making* contribute to this world of shared, informed decision making using computer-based decision analysis? As in the past, textbooks will shape the way that future decision analysts learn and later practice the discipline of their life's work. We think that clinicians-in-training should master the material in Chapters 3, 4, and 5. They should be able to use the time-tradeoff method to assess a patient's utilities for a health state. They should have a cultural understanding of decision modeling. Finally, we hope that aspiring master clinicians will read a book like ours to gain a greater understanding of their daily work and the limitations of the imperfect information that they rely upon.

H.C.S.

Introduction

“Proof,” I said, “is always a relative thing. It’s an overwhelming balance of probabilities. And that’s a matter of how they strike you.”

(Raymond Chandler, Farewell, My Lovely, 1940)

Probability is the rule of life – especially under the skin. Never make a positive diagnosis.

(Sir William Osler)

Thoughtful clinicians ask themselves many difficult questions during the course of taking care of patients. Some of these questions are as follows:

- How may I be thorough yet efficient when considering the possible causes of my patient’s problem?
- How do I characterize the information I have gathered during the medical interview and physical examination?
- How should I interpret new diagnostic information?
- How do I select the appropriate diagnostic test?
- How do I choose among several risky treatments?

The goal of this book is to help clinicians answer these important questions.

The first question is addressed with observations from expert clinicians “thinking out loud” as they work their way through a clinical problem. The last four are addressed from the perspective of medical decision analysis, a quantitative approach to medical decision making.

The goal of this introductory chapter is to preview the contents of the book by sketching out preliminary answers to these five questions.

1.1 How may I be thorough yet efficient when considering the possible causes of my patient’s problems?

Trying to be efficient in thinking about the possible causes of a patient’s problem often conflicts with being thorough. This conflict has no single solution. However, much may be learned about medical problem solving

by listening to expert diagnosticians discuss how they reasoned their way through a case. Because the single most powerful predictor of skill in diagnosis is exposure to patients, the best advice is “see lots of patients and learn from your mistakes.” How to be thorough, yet efficient, when thinking about the possible causes of a patient’s problem is the topic of Chapter 2.

1.2 How do I characterize the information I have gathered during the medical interview and physical examination?

The first step toward understanding how to characterize the information one gathers from the medical interview and physical examination is to realize that information provided by the patient and by diagnostic tests usually does not reveal the patient’s true state. A patient’s signs, symptoms, and diagnostic test results are usually representative of more than one disease. Therefore, distinguishing among the possibilities with absolute certainty is not possible. A 60-year-old man’s history of chest pain illustrates this point:

Mr. Costin, a 60-year-old bank executive, walks into the emergency room complaining of intermittent substernal chest pain that is “squeezing” in character. The chest pain is occasionally brought on by exertion but usually occurs without provocation. When it occurs, the patient lies down for a few minutes, and the pain usually subsides in about 5 minutes. It never lasts more than 10 minutes. Until these episodes of chest pain began 3 weeks ago, the patient had been in good health, except for intermittent problems with heartburn after a heavy meal.

Although there are at least 60 causes of chest pain, Mr. Costin’s medical history narrows down the diagnostic possibilities considerably. Based on his history, the two most likely causes of Mr. Costin’s chest pain are coronary artery disease or esophageal disease.

However, the cause of Mr. Costin’s illness is uncertain. This uncertainty is not a shortcoming of the clinician who gathered the information; rather, it reflects the uncertainty inherent in the information provided by Mr. Costin. Like most patients, his true disease state is hidden within his body and must be inferred from imperfect external clues.

How do clinicians usually characterize the uncertainty inherent in medical information? Most clinicians use words such as “probably” or “possibly” to characterize this uncertainty. However, most of these words are imprecise, as illustrated as we hear more about Mr. Costin’s story:

The clinician who sees Mr. Costin in the emergency room tells Mr. Costin, “I cannot rule out coronary artery disease. The next step in the diagnostic process is to examine the results of a stress ECG.” She also says, “I cannot rule out esophageal disease either. If the stress ECG is negative, we will work you up for esophageal disease.”

Mr. Costin is very concerned about his condition and seeks a second opinion. The second clinician who sees Mr. Costin agrees that coronary artery

disease and esophageal disease are the most likely diagnoses. He tells Mr. Costin, “Coronary artery disease is a likely diagnosis, but to know for certain we’ll have to see the results of a stress ECG.” Concerning esophageal disease, he says, “We cannot rule out esophageal disease at this point. If the stress ECG is normal, and you don’t begin to feel better, we’ll work you up for esophageal disease.”

Mr. Costin feels reassured that both clinicians seem to agree on the possibility of esophageal disease, since both have said that they cannot rule it out. However, Mr. Costin cannot reconcile the different statements concerning the likelihood that he has coronary artery disease. Recall that the first clinician said “coronary artery disease cannot be ruled out,” whereas the second clinician stated, “coronary artery disease is a likely diagnosis.” Mr. Costin wants to know the difference between these two different opinions. He explains his confusion to the second clinician and asks him to speak to the first clinician:

The two clinicians confer by telephone. Although the clinicians expressed the likelihood of coronary artery disease differently when they talked with Mr. Costin, it turns out that they had similar ideas about the likelihood that he has coronary artery disease. Both clinicians believe that about one patient out of three with Mr. Costin’s history has coronary artery disease.

From this episode, Mr. Costin learns that clinicians may choose different words to express the same judgment about the likelihood of an uncertain event:

To Mr. Costin’s surprise, the clinicians have different opinions about the likelihood of esophageal disease, despite the fact that both clinicians described its likelihood with the same phrase, “esophageal disease cannot be ruled out.” The first clinician believes that among patients with Mr. Costin’s symptoms, only one patient in ten would have esophageal disease. However, the second clinician thinks that as many as one patient in two would have esophageal disease.

Mr. Costin is chagrined that both clinicians used the same phrase, “cannot be ruled out,” to describe two different likelihoods. He learns that clinicians commonly use the same words to express different judgments about the likelihood of an event.

The solution to the confusion that can occur when using words to characterize uncertainty with words is to use a number: probability. Probability expresses uncertainty precisely because it is the likelihood that a condition is present or will occur in the future. When one clinician believes the probability that a patient has coronary artery disease is 1 in 10, and the other clinician thinks that it is 1 in 2, the two clinicians know that they disagree and that they must talk about why their interpretations are so disparate. The precision of numbers to express uncertainty is illustrated graphically by the scale in Figure 1.1. On this scale, uncertain events are expressed with numbers between 0 and 1.

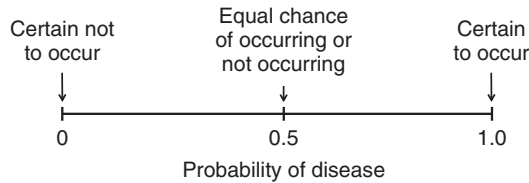


Figure 1.1 A scale for expressing uncertainty.

To understand the meaning of probability in medicine, think of it as a fraction. For example, the fraction “one-third” means 33 out of a group of 100. In medicine, if a clinician states that the probability that a disease is present is 33%, it means that the clinician believes that if she sees 100 patients with the same findings, 33 of them will have the disease in question (Figure 1.2).

Although probability has a precise mathematical meaning, a probability estimate need not correspond to a physical reality, such as the prevalence of disease in a defined group of patients. We define probability in medicine as a number between 0 and 1 that expresses a clinician’s opinion about the likelihood of a condition being present or occurring in the future. The probability of an event a clinician believes is certain to occur is equal to 1. The probability of an event a clinician believes is certain not to occur is equal to 0.

A probability may apply to the present state of the patient (e.g., that he has coronary artery disease), or it may be used to express the likelihood that an event will occur in the future (e.g., that he will experience a myocardial infarction within one year).

Any degree of uncertainty may be expressed on this scale. Note that uncertain events are expressed with numbers between 0 and 1. Both ends of the scale correspond to absolute certainty. An event that is certain to occur is expressed with a probability equal to 1. An event that is certain not to occur is expressed with a probability equal to 0.

When should a clinician use probability in the diagnostic process? The first time that probability is useful in the diagnostic process is when the clinician feels she needs to synthesize the medical information she has obtained in the medical interview and physical examination into an opinion. At this juncture the clinician wants to be precise about the uncertainty because she is poised

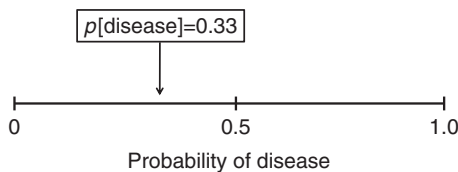


Figure 1.2 A clinician can visualize the level of certainty about a disease hypothesis on a probability scale. Thirty-three is marked on this certainty scale to correspond to the clinician’s initial probability estimate concerning the likelihood that Mr. Costin had coronary artery disease.

to make decisions about the patient. The clinician may decide to act as if the patient is not diseased. She may decide that she needs more information and will order a diagnostic test. She may decide that she knows enough to start the patient on a specific treatment. To decide between these options, she does not need to know the diagnosis. She does need to estimate the patient's probability that he has, as in the case of Mr. Costin, coronary artery disease as the cause of his chest pain.

A clinician arrives at a probability estimate for a disease hypothesis by using personal experience and the published literature. Advice on how to estimate probability is found in Chapter 3.

1.3 How do I interpret new diagnostic information?

New diagnostic information often does not reveal the patient's true state, and the best a clinician can do is to estimate how much the new information has changed her uncertainty about it. This task is difficult if one is describing uncertainty with words. However, if the clinician is expressing uncertainty with probability, she can use Bayes' theorem to estimate how much her uncertainty about a patient's true state should have changed. To use Bayes' theorem a clinician must estimate the probability of disease before the new information was gathered (the *prior probability* or pre-test probability) and know the accuracy of the new diagnostic information. The probability of disease that results from interpreting new diagnostic information is called the *posterior probability* (or post-test probability). These two probabilities are illustrated in Figure 1.3.

Chapter 4 describes how to use Bayes' theorem to estimate the post-test probability of a disease.

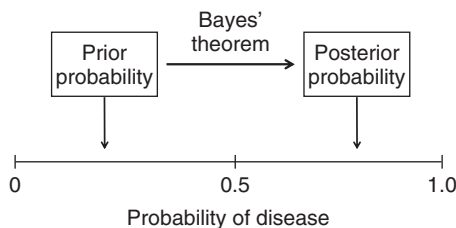


Figure 1.3 The pre-test probability and the post-test probability of disease.

1.4 How do I select the appropriate diagnostic test?

Although the selection of a diagnostic test is ostensibly straightforward, the reasoning must take into account several factors. In the language of medical decision analysis, the selection of diagnostic tests depends on the patient's feelings about states of disease and health, the clinician's estimate of the prior probability of disease, and the accuracy of the diagnostic tests that the clinician is trying to choose between.

A logical approach to selecting diagnostic tests depends on three principles:

- Diagnostic tests are imperfect and therefore seldom reveal a patient's true state with certainty.
- Tests should be chosen if the results could change the clinician's mind about what to do for the patient.
- Clinicians often start treatment when they are uncertain about the true state of the patient.

These three principles lead to an important concept: The selection of diagnostic tests depends on the level of certainty at which a clinician is willing to start treatment. This level of certainty is known as the treatment-threshold probability. How to use the treatment-threshold probability to make decisions is a topic of Chapter 9.

A clinician must take two steps to assess the treatment-threshold probability of disease. The first step is to list the harms and benefits of treatment. The second step is to assess the patient's feelings about these harms and benefits. A decision analyst assesses a patient's attitudes toward the risks and benefits of treatment using a unit of measure called *utility*. Measuring a patient's utilities is covered in Chapter 8 of this text.

1.5 How do I choose among several risky treatment alternatives?

Choosing among risky treatment alternatives is difficult because the outcome of most treatments is uncertain: some people respond to treatment but others do not. If the outcome of a treatment is governed by chance, a clinician cannot know in advance which outcome of the treatment will result. Under these circumstances, the best way to achieve a good outcome is to choose the treatment alternative whose average outcome is best. This concept is called *expected value decision making*. Expected value decision making is the topic of Chapters 6, 7, and 11.

1.6 Summary

The care of patients is difficult in part because of the uncertainty inherent in the nature of medical information: tests are imperfect, and treatments have unpredictable consequences. The application of probability, utility, and expected value decision making provides a framework for making the right decision despite the uncertainty of medical practice. Medical decision analysis helps clinicians and patients to cope with uncertainty.

Differential diagnosis

This chapter is about differential diagnosis, a systematic process for narrowing the range of possible explanations for a patient's problem. The goal of this chapter is to describe a thorough, yet efficient, approach to this process. The chapter has four parts:

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2.1 Introduction

Differential diagnosis is the process of considering the possible causes of a patient's symptom or physical finding and making a diagnosis. Differential diagnosis is a safeguard against premature conclusions as well as a time-proven method for attacking what can be a supremely difficult intellectual challenge. All clinicians, regardless of their specialty, use differential diagnosis and strive to master it. For many clinicians, to be called a superb diagnostician is the highest form of praise.

The Challenge of Differential Diagnosis: A patient visits your office on a busy afternoon because of a symptom. She wants you to discover the reason for the symptom and then cure it quickly and painlessly. You must discover its cause or at least assure yourself that it is not due to a serious, treatable disease. This task can be difficult, especially when the symptom has many possible causes. It is hard to recall a long list of possible causes or the key features of even one cause of the symptom. Moreover, these key features typically occur in more than one disease. Time pressure often adds to the intellectual difficulties.

The Purposes of the Interview: The medical interview has many purposes. The first goal is to establish a relationship of mutual trust with the patient. Another goal is to observe the patient closely for clues that may help to focus your investigation. A third goal is to narrow the list of diseases that could be causing the patient's problem and focus the physical examination on a few possible diagnoses. Later on comes the physical examination and decisions about whether to perform diagnostic tests.

2.2 How clinicians make a diagnosis

We know more about how clinicians *should* reason than about how they *do* reason. This book is about how physicians should reason in specific situations, such as selecting and interpreting diagnostic tests or making a decision between two treatments. These normative methods are based on first principles of logical reasoning and take full advantage of all sources of information. This chapter is about how physicians *do* reason as they strive to narrow the list of possible causes of a patient's symptom.

This chapter describes a logical framework for diagnosis, but we cannot claim that it is based on first principles of logical reasoning. We can claim that expert clinicians use the process. However, the path to expertise only begins with a good process. Research shows that expert clinicians got that way by seeing lots of patients. Moreover, they are skillful at applying what they learned from past experience to solve the problem of the moment.

The process that we will describe is based upon listening to expert diagnosticians think aloud as they solve a diagnostic problem. Diagnosticians, expert or not, use a similar process. The expert has a larger, more accessible fund of knowledge than the average clinician. Some of this knowledge comes from textbooks and articles, but most of it comes from having seen similar patients and remembering their diagnosis (pattern recognition).

The message for beginning students is two-fold. First, they will learn a process that is the foundation of excellence in diagnosis. Second, the pathway to excellence is open to those who constantly strive to enlarge their experience by immersing themselves in clinical medicine and the important details of individual patients and their ultimate fate. The more you challenge yourself, the more you will learn and the more expert you will become. You do not get better by playing it safe and avoiding exposure to the possibility of being wrong. In fact, experts will tell you that they learn more from being wrong than from being right.

The central mystery of medical education is how students acquire skill in differential diagnosis. This topic is seldom taught as a formal discipline. Most learn it while discussing specific patients with a clinical teacher. This gap in the medical curriculum is easily understood: our understanding of the methods used by skilled diagnosticians would scarcely sustain an hour's lecture. What we do know about differential diagnosis is the result of research in which clinicians "think out loud" as they work their way through a diagnostic problem. These observations may be summarized in five conclusions:

1. Hypotheses are generated early in the interview.
2. Only a few hypotheses are being actively considered at any moment.
3. Newly acquired information is often used incorrectly.
4. Clinicians often fail to take full advantage of past experience with similar patients.
5. A clinician's skill in differential diagnosis varies from topic to topic; more experience usually means greater expertise.

Each one of these conclusions requires additional comment:

1. *Hypotheses are generated early:* Beginning students are often instructed to obtain all relevant data before starting to exclude diseases from the list of possible causes of the patient's complaint. They soon outgrow this mistaken teaching. Experienced clinicians observe the patient closely while introducing themselves and often begin to draw tentative conclusions before the patient has spoken. As soon as they identify the patient's main complaint, they use the patient's age and gender to help identify the main diagnostic possibilities. Good diagnosticians will ask the patient to tell the story of the main complaint while they listen, observe, and formulate hypotheses. These hypotheses determine their first specific questions, and the answers bring other hypotheses to mind.

Conclusion: data collection is hypothesis-driven.

2. *Only a few hypotheses are considered simultaneously:* Research about cognition shows that the human mind has a limited working memory. Just as a juggler can keep only a few objects in the air at once, so the clinician can consider only a few hypotheses simultaneously.

The clinician begins evaluating a hypothesis by matching the patient's findings with the clinician's internal representation, or model, of the disease. Sometimes this internal representation is the pathophysiology of the condition. One pathophysiological derangement causes another, as inferred by logical deduction, which leads to another and eventually to the expected clinical manifestations. Reasoning backward along this chain of logical deductions may lead to the cause of the patient's problem. One way to remember the differential diagnosis of a low serum sodium is to reason deductively from the pathophysiology of the condition. Research indicates that expert diagnosticians use pathophysiological reasoning mainly for difficult, atypical cases. Expert diagnosticians typically have an excellent understanding of pathophysiology.

More often, the clinician uses an associative model of disease, also known as pattern recognition. Associative models consist of clinical findings, illness progression, predisposing characteristics, and complications that are associated with a disease. The clinician asks about the typical features of a disease and is often able to eliminate hypotheses solely from the patient's responses. Hypotheses that cannot be readily discarded are further tested during the physical examination. Another mark of an expert diagnostician is the ability to quickly match the patient's findings to a pattern of disease manifestations. Often, the patient's findings remind the clinician of a similar patient seen many years earlier. Expert clinicians retain these experiences and can recall specific patients from many decades earlier.

The supreme mystery of clinical reasoning is the cognitive process that clinicians use to discard or confirm a hypothesis. Several models of this process have been proposed. These models are based on analyzing what clinicians say about how they test hypotheses:

- *Additive model*: In a linear model, clinicians assign a positive weight to findings that tend to confirm a diagnosis and negative weights to findings that disconfirm the diagnosis. In some unknown cognitive process, the clinician decides to discard or accept a hypothesis based on the sum of the diagnostic weights. Clinical prediction rules use this model (see Chapter 3). Physicians probably assign weights subjectively and subconsciously. In the context of this chapter, the analog of the diagnostic weight of a finding might be the likelihood ratio of the finding. The likelihood ratio indicates whether the odds of the target condition go up or down and by how much. It is, therefore, an empirical, quantitative measure of the changes in the odds of the target condition (see Chapters 4 and 5).
- *Bayesian model*: Bayes' theorem is the method to calculate the post-test probability of the target condition. Bayes' theorem tells the clinician how much new information should change the probability. By analogy, clinicians may change their belief in a hypothesis with each new item of information and conclude at some point that the probability is low enough to discard the hypothesis.
- *Algorithmic model*: Algorithms are commonly used to represent the logic of diagnosis. Do clinicians follow an internal flow sheet with branching logic as they test a hypothesis? Does a series of "no" branches eventually lead to discarding a hypothesis? We do not know.

Undoubtedly, clinicians use features of these admittedly speculative models as they consider a diagnostic hypothesis, but the essence of diagnostic reasoning eludes understanding.

3. *Newly acquired information is often used inappropriately*: Mistakes happen when a clinician begins to believe in a diagnostic hypothesis. The clinician may ignore conflicting information or misinterpret it as confirming an existing hypothesis when the correct action is to disregard it, use it to reject the hypothesis, or use it as a clue to a new hypothesis. The clinician may exaggerate the importance of findings that fit with a preconceived idea or accommodate inconsistent data by reformulating a hypothesis to the point where it is too general to be tested parsimoniously.

Example: These errors are illustrated by the story of the medical student who offered a diagnosis of leishmaniasis in a patient with diffuse lymphadenopathy. The patient had taken a steamship cruise to a South American port years before. When the attending physician asked him to justify this arcane diagnosis, the student replied, "what else causes lymphadenopathy?" Unable to remember any other causes of lymphadenopathy, the student exaggerated the importance of a rather dubious travel history in order to support a far-out hypothesis.

Conclusion: placing too much weight on data to support a low-probability hypothesis leads to mistakes.

4. *Past experience can be used inappropriately:* Clinicians use rules of thumb (heuristics) for using experience to estimate probability from the match between the patient's case history and past experience. These heuristics include representativeness (the goodness of fit between the patient's case and past cases) and availability (how easily past cases come to mind). Thoughtless use of these heuristics can lead to mistakes (discussed at length in Chapter 3).

5. *Skill in clinical reasoning varies from topic to topic:* Students of cognitive psychology have assumed that individuals who are able to reason well about one topic should be equally proficient with other topics. In fact, clinicians' reasoning skills vary from topic to topic. Associations between hypotheses and clinical findings are learned by experience. Therefore, mastery of a topic requires experience with a wide variety of cases. A good memory and cognitive facility are not enough.

Conclusion: expertise requires a deep clinical experience.

Summary: Descriptive studies of clinical reasoning provide helpful insights for the beginning clinician. However, despite dogged efforts by researchers, we do not understand many aspects of clinical reasoning.

2.3 The principles of hypothesis-driven differential diagnosis

The most important conclusion from research on how experts make a diagnosis is that the questions they are asking at any moment are intended to test the small number of diagnostic hypotheses that they are considering at that moment. In other words, the main function of the interview and physical examination is to test diagnostic hypotheses. Thinking of this principle may help the clinician to steer the interview back on topic when it threatens to get sidetracked.

While reading this chapter, imagine that you are evaluating a patient with a chief complaint of "nocturnal chest pain for one month." The pain is a substernal tightness that begins an hour or so after lying down. A complete evaluation of this complaint will include the following steps:

1. Taking the history.
2. Doing a physical examination.
3. Selecting and interpreting diagnostic tests.
4. Choosing a treatment.

This book will cover the principles underlying each of these steps in considerable detail. The purpose of this chapter is to describe the thinking processes that guide the history and physical examination. Subsequent chapters prepare the reader to choose diagnostic tests and therapy.

Differential diagnosis is a cyclic process consisting of three steps which ultimately lead to a fourth step, taking action (Figure 2.1).

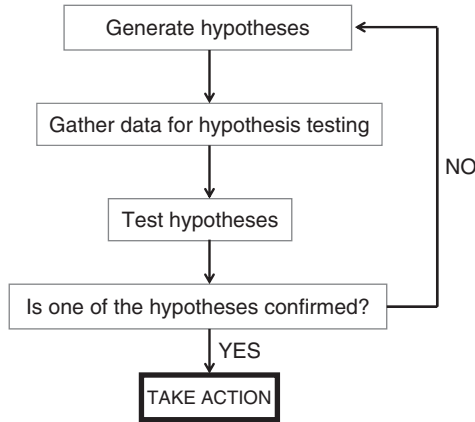


Figure 2.1 The cyclic process of differential diagnosis.

The three steps are repeated many times as hypotheses are considered and rejected, confirmed, or set aside for further testing. Steps 2 and 3 are repeated during the physical examination. The following four sections follow the cyclic process shown in Figure 2.1.

2.3.1 The first step in differential diagnosis: listening and generating hypotheses

This process begins as the patient voices the chief complaint. Most clinicians ask the patient to provide a chronologic account of his illness from its beginning. This approach provides valuable information and perspective on the patient’s illness. It also respects the patient. As the patient tells his story, the clinician has time to think, write down some diagnoses to consider, and observe the patient for diagnostic clues.

Hypothesis generation begins at the start of the interview. The first catalyst for hypothesis generation is usually the chief complaint, although the patient’s appearance and agility as he take his seat in the examining room also provide a context for interpreting the history. As shown in Figure 2.2, the

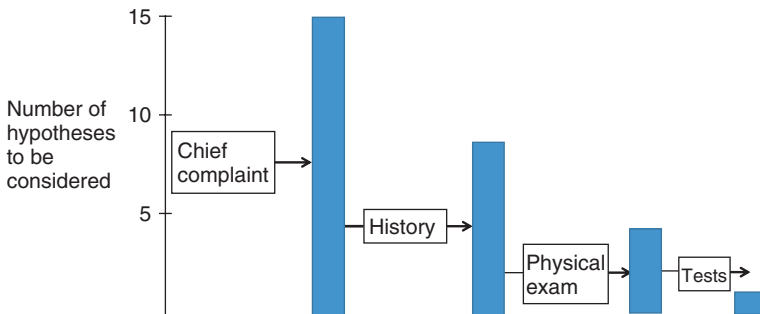


Figure 2.2 Number of diagnostic hypotheses remaining during the steps of evaluating a symptom.