Editorial

Every first-year medical student learns that diagnosis must precede treatment, but few are taught to acknowledge their own fallibility or to manage the uncertainty that plagues every decision. Read on to understand the perils of denying diagnostic uncertainty and what can be done to act intelligently when it inevitably surfaces.

Letter to the Editor

Why is it that almost all medical news deals with treatment and rarely, if ever, mentions diagnosis? I suppose that hope springs eternal and that little is gained from making a proper diagnosis when there is an endless abundance of drugs, interventions, and surgical procedures to employ with boundless optimism. Sooner or later something works, and, if not, a placebo effect or spontaneous resolution will save the day. Since most common ailments have their ups and downs, and patients seek care when down, they are likely to head upwards after a doctor’s visit (regardless of what is done), courtesy of a statistical phenomenon called “regression to the mean.”

Maybe diagnosis is woefully overrated: with more healing power than ever before, why waste time being accurate when something close will likely suffice? Paradoxically, there have been similar advances in diagnostic tests and imaging studies, yet new technology can hinder proper diagnosis and delay helpful treatment.

What can be done to restore good old-fashioned detective work to medical encounters, whereby the doctor listens carefully, hears the patient’s story, and thoughtfully puts the pieces together into a clear diagnosis before plunging headfirst into treatment? I look forward to learning your thoughts on this ever important issue.

Diagnostically yours,

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Editor’s Response

If you want to get your doctor’s attention, ask two simple questions: (1) what’s your diagnosis? and (2) how certain are you? The second is likely to get the stronger reaction, since implicitly it suggests she may be wrong or, at the very least, not fully certain of the verdict. And if the first two questions did not provoke enough of a reaction, ask her if she washed her hands; but that’s another story.

So why obsess over your doctor’s diagnostic certainty? Because, quite simply, everything else depends on it. Without proper diagnosis, even the most adroit application of costly cutting-edge therapy will not produce the desired benefit and may very well result in unanticipated harms or adverse events. As Anna Walker cautioned, “It’s very important that patients and the public understand the benefits of treatment and follow-up care—and the risks that will result without diagnosis.”

If you are a parent, consider that last time you showed up at the doctor when your child had fever and sore throat. Although almost certainly a viral infection, an antibiotic may have been inappropriately prescribed, especially if the doctor was older or very busy. Ditto for childhood ear infections, for which diagnosis can be difficult yet antibiotics are given despite uncertainty. Whereas both conditions just mentioned are usually self-limited, coronary heart disease is not, yet it is still plagued by practice variations related to diagnostic certainty: US physicians are most certain (not surprisingly) and German physicians the least.

A dirty little secret in medicine is that doctors are often uncertain but rarely admit it. Regrettably, making a certain diagnosis can be incredibly difficult; a patient’s signs, symptoms, and test results rarely fall into the neat diagnostic patterns in textbooks, and in all but the most straightforward cases, there is always—and I repeat always—an inevitable core of uncertainty. Uncertainty is not necessarily bad when recognized and appropriately managed. Truth takes time to operate, and so can diagnosis; what is not apparent at first encounter may gradually emerge as follow-up adds nuance and clarity.

Jerome Groopman, who highlights the issue of diagnostic certainty in How Doctors Think, attributes 3 basic types of uncertainty to Renee Fox: imperfect mastery of knowledge, limitations in available knowledge, and the difficulty in distinguishing among the first two. I suspect that the last two underlie most medical uncertainty, since doctors can access existing knowledge with relentless efficiency using online and point-of-care resources. Groopman observes that “specialists in particular are known to demonstrate unwarranted clinical certainty. They have trained for so long that they begin to easily rely on their vast knowledge and overlook the variability in human biology.”

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The trick to better diagnosis is to admit uncertainty, acknowledge it when present, and take steps to minimize it once identified. Regardless of your presumed level of medical savvy, consider the following before accepting a doctor’s proclamation as the gospel:

1. Assess your doctor’s listening skills. All medical diagnosis begins with history taking. A sharp clinician listens attentively, interrupts only when necessary, and pays as much attention to the patient’s narrative as the facts related. Effective medical practice requires narrative competence: an ability to absorb, interpret, and act on the stories and plights of others.7 As the saying goes, we have two ears and one mouth for good reason: listening should be twice as important. The average clinician interrupts patients within seconds of their story; if this is your doctor, think carefully about your confidence in his or her listening skills.

2. Pay careful attention to the examination. Some diagnoses are made almost exclusive by symptoms, but most require confirmatory physical findings. The diligence—or lack thereof—given to examination can sharpen diagnostic accuracy or lead the clinician astray. Unless you are a physician yourself, your best guide to the adequacy of examination is often a gut feeling: was the doctor fully engaged or did he or she seem rushed, cursory, or too narrowly focused? There’s nothing wrong with explicitly asking, “So what exactly did you find on your exam?” if you want more information.

3. Be a diagnostic skeptic. A diagnosis, or range of diagnoses, is usually rendered after interviewing and examining the patient. But remember there are always two possibilities: the doctor is right, or the doctor is wrong. The situation is more complicated since doctors may think they are right when really wrong or think they are wrong when really right. A sharp clinician thinks in terms of probabilities, not just admitting the possibility of error but estimating the likelihood. The only “certainty” in medicine is delusory certainty: a conceited self-importance that raises someone above inevitable errors in judgment. As the recipient of a medical diagnosis, be sure to ask, “How sure are you?” and “Are there other possibilities?”

4. Question all diagnostic tests. Tests are appropriate when their ability to reduce uncertainty more than offsets any attendant risks, harms, and costs. Clinicians should explicitly discuss these issues, and patients are entitled to a meaningful explanation. An attitude of “well it can’t hurt to do more tests” should be patently avoided, since false-positive results do occur and can lead to unnecessary biopsies, surgery, or procedures. When a test is advised, be sure to ask, “How accurate is this test, what’s the downside, and how could it alter management?”

5. Get a second opinion. If an illness is brief, mild, and uncomplicated, the effort in getting a second opinion is rarely warranted. Conversely, a second opinion is prudent when treatment has significant side effects, the problem persists or recurs despite treatment, or the problem is complex (rare diseases, multiple chronic conditions) or progressive (cancer, neuromuscular disorder). A timely second opinion can reduce uncertainty and boost confidence in a management plan. Letting your doctor know about a second opinion can be revealing; if he or she shuns the prospect or seems annoyed, you may need to look elsewhere.

We can feel most confident about a diagnosis when accompanied by a diligent medical history, an adequate physical examination, a credible statement of confidence (eg, unsure, possibly, very likely, almost certain), a brief statement of other possibilities, and a comment about the need, if any, for further tests. Most important, there should be willingness to accept the possibility of being wrong. Last, there should be a clear plan for future efforts to confirm, or reassess, the diagnosis should the condition worsen or not respond as anticipated.

The scope of diagnostic tests available to clinicians increases daily, fueled by new technology that aids and abets a seductive quest for certainty. An ideal test would correctly identify 100% of individuals with a condition (sensitivity) and 100% of those without it (specificity). Some tests may come close (so-called gold standards), while others may be no better than a coin toss. Most fall somewhere in between, excelling at either sensitivity or specificity, but much less often at both. On the basis of these properties, you should ask for the accuracy (or predictive value) of a positive result and a negative one, either of which could potentially be wrong.

Test results are commonly categorized as “positive” or “negative,” conveniently overlooking the possibility of an “uncertain” outcome.8 Failing to acknowledge uncertainty in a positive result could delay proper treatment or result in unnecessary complications or side effects. Failing to acknowledge uncertainty in a negative result could instill false confidence, allowing a serious, but undiagnosed, condition to progress. The answer is not simply more tests, since each carries its own risk of error that multiplies prior uncertainty. Only by admitting uncertainty can a measured and thoughtful solution arise to its reduction.

I have written previously on the inevitable uncertainty in medical practice, so why dwell on it again when writing about diagnosis? Because effective medical care rests on a platform of sound diagnosis, which then sets the stage for counseling, ordering tests, considering natural history, and weighing options for management. A wrong diagnosis in medicine is akin to management without leadership in business: expert implementation of a flawed idea or vision rarely produces a desired outcome unless luck intervenes. I would humbly suggest that “luck” is not the ideal platform on which to condition a medical outcome.
Modern medicine could benefit from some philosophy or, more specifically, epistemology: studying the nature and grounds of knowledge, especially with reference to its limits and validity. We might begin this task by confessing our limits of medical knowledge, by accepting the accompanying uncertainty, and by conducting research on how we can best manage whatever inevitable uncertainty exists. Some commonsense management strategies include confessing uncertainty to patients, encouraging second opinions, and referring the patient to another clinician with more relevant expertise when appropriate. A less useful approach is to sidestep the issue of diagnosis by simply assigning nonspecific medical terms to the patient’s symptoms (eg, dysphonia for hoarseness, pharyngitis for sore throat, rhinitis for runny nose).

Diagnosis is ultimately pattern recognition: finding the specific illness, disorder, or syndrome that best accounts for the signs, symptoms, and test results. Most often, however, the image is more akin to an impressionist painting than photographic realism. But even the fuzzy shapes of impressionism assume new clarity when viewed from afar, just as a fuzzy diagnosis can achieve clarity with thought, reflection, and a new viewpoint or a second opinion. And sometimes, of course, diagnosis remains elusive despite best efforts. This brings to mind Osler’s advice: “To confess ignorance is often wiser than to beat about the bush with a hypothetical diagnosis.”

Granted this suggestion is more than a century old, but admitting our own fallibility will likely never go out of style.

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References