Stumbling towards a diagnosis

Abstract: Not all diagnostic successes occur easily or as flashes of inspiration. Sometime the key feature in diagnostic success is persistence.

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I remember hearing the phrase “great diagnostician” in my youth. Patients ascribed this description to physicians they admired. Diagnosticians were the most desired physicians. Sometime over the past 40 years the importance of diagnostic acumen seemedly decreased. We worry about treatment algorithms and process measures of performance and accurate coding rather than worry about diagnosis. While medical education seems to have de-emphasized diagnosis, and “quality report cards” ignore diagnosis, our patients still want a correct diagnosis. Patients appreciate our striving to make a firm diagnosis.

During my career, nothing has captured my attention more that striving to make a diagnosis. As a medical detective I want to solve the mystery. Sometimes I succeed. But in considering my detective process, I realize that while occasionally I have a moment of inspiration, more often I am stumbling towards a diagnosis, and that description is both apt and acceptable.

As I have considered this concept, I have thought about the influence that mystery books and television shows have had on my understanding of the diagnostic process. Two quotes help frame the process. The first quote comes, not surprisingly, from Sherlock Holmes, not from the written canon, but rather from the brilliant BBC series – Sherlock. He comes to a crime scene where a rookie detective declares that he has already solved the crime. Sherlock points out a few inconsistencies that the rookie ignored. “You have a solution that you like, but you are choosing to ignore anything that you see that doesn’t comply with it.” When stumbling towards a diagnosis, one must not ignore any clues, even if those clues seem inconvenient.

For years Columbo has fascinated me. His classic head scratching inquisitive questioning says a great deal about the diagnostic process. He would be questioning a suspect and, “Oh, excuse me, sir, but just one more question. I’ been thinkin’ and it strikes me kinda funny that ...” He seemed puzzled, but he was always working to evaluate his hypothesis.

The field of naturalistic decision-making focuses on the development and characterization of expertise. How does one become an expert? These psychologists observe experts as they make decisions; especially under time-constrained high stakes situations. In their studies they find that experts evaluate possibilities one at a time. They use system 1 thinking (the experiential or pattern recognition process) to identify a potential diagnosis. They then test this diagnosis against the facts, both those facts present and what additional facts they need to actually feel confident of their diagnosis.

The classic television mystery series use this approach. Recently I have watched several episodes of “The Glades”. In this show, as each new clue is discovered, the main character – Jim Longworth – interviews the suspect. Something in the interview either excludes that suspect or at least makes the sequence of presumed events unlikely. He and his team then uncover another clue, pointing to another suspect. The process repeats, until they discover the final clue and confront the perpetrator armed with the evidence.

In this show, and many other similar shows, the detective is actually stumbling. But stumbling is not bumbling. Stumbling is a good description of the diagnostic process. Stumbling implies continuing our search for a diagnosis. Stumbling avoids premature closure, diagnostic momentum and other diagnostic evils. Stumbling in this use means intelligent persistence and knowing that the clues do not yet “add up”.

We saw a patient recently that epitomizes the stumbling paradigm. He was in his early forties, Hispanic and had lived in the United States for approximately 10 years, who presented with increasing dyspnea and fever episodes. He had a mild cough of clear sputum with occasional blood streaks.

His initial chest X-ray showed bilateral infiltrates, right greater than left. The admitting physicians assumed that he had community-acquired pneumonia and thus started routine antibiotics. Over the following 3 days his fever and dyspnea persisted. Both infectious disease and pulmonary consults suggested changing his antibiotic coverage to include less frequent pneumonia causes.
During this time the physicians caring for him frequently expressed concern, as the patient’s appearance did not suggest a severe pneumonia. While they prescribed antibiotics, they kept questioning the diagnosis.

On day 8 he developed worsening hemoptysis. His hemoglobin decreased significantly. Tests for tuberculosis returned negative.

On day 10 his chest X-ray worsened on the left, but had improved slightly in the right lung. A bronchoscopy showed diffuse alveolar hemorrhage.

The team then reassessed his presentation and reviewed his tests. His initial creatinine of 0.8 mg/dL had increased to 1.2 mg/dL. They discovered that he had not had a urinalysis, and the analysis that day showed 180 RBC per high power field without casts.

Rethinking the patient presentation provided two major new suspects. We had now decided that this patient neither had community acquired pneumonia or a more complex bacterial pneumonia. While we lamented the lack of an initial urinalysis, we now focused on pulmonary renal syndromes. We sent laboratory testing for the two most likely suspects – granulomatous polyangiitis and Goodpasture’s syndrome. Fortunately, the initial treatment for both included exchange transfusions and high dose corticosteroids. Our renal consult also performed a renal biopsy to guide renal disease management.

On day 13 we finally identified the guilty suspect. The biopsy showed crescentic glomerulonephritis and the blood tests showed cANCA (positive anti-neutrophil cytoplasmic antibodies) positive as well as pR3Ab, confirming the culprit disease as granulomatosis with polyangiitis (formerly known as Wegener’s granulomatosis).

Our diagnostic pathway was indirect, tortuous and confusing. We kept testing hypotheses until we finally found the diagnosis that explained all the findings.

Successful stumbling requires two attributes. First, one must recognize diagnostic discomfort and avoid diagnostic momentum. Repeatedly the physician team expressed their discomfort concerning the diagnosis. As one of our colleagues often says, the team dated a diagnosis, but felt the diagnosis did not meet marriage criteria.

Second, one must continue searching for new clues. Stumbling is a conscious process. It requires recognition of uneasiness with the “working” diagnosis.

When we realized that the patient’s presentation did not fit the initial diagnosis, then we rebooted the diagnostic process.

We stumbled, but stumbling is not necessarily bad. Sometimes we receive diagnostic inspiration, but more often we continue searching doggedly. As long as we continue revisiting the story and the available tests, our stumbling has a good chance to fall upon the correct diagnosis. The important concept is persistence to confirm a correct diagnosis. Stumbling, while not elegant, beats diagnostic arrogance every time.

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