Why is Diagnosing So Hard?

By Ann F. Beach, MD, FAAP

The correct diagnosis is crucial. If we don't get the right diagnosis, we can't treat and cure our patients.

However, getting to that correct diagnosis can be devilishly difficult. It is a constant struggle for us doctors and requires knowledge, patience, experience, persistence, the help of colleagues and, sometimes, a sixth-sense. (I call it the spidey-sense.)

To add to the difficulty, the initial diagnosis is not always the true diagnosis. In fact, 30% of patients surveyed say that the initial diagnosis they were given was not ultimately their final diagnosis.

It's not what anyone wants. Doctors work hard to get the right diagnosis from the start, because the right diagnosis leads to the right treatment.

So why is diagnosis hard? There are a limited number of problems a patient may show up with (rash, fever, fatigue, cough), yet each of these symptoms can represent thousands of diagnoses. There are, after all, over 10,000 recognized diseases, and that number is constantly growing.

Think of the patient's presenting complaint as a tree trunk, and the thousands of branches and twigs being the possible diagnoses. So a nagging cough is likely to be from a cold, but it could be pneumonia, a small piece of food aspirated into the lung, a lung tumor … or a rare bat-borne infection in a spelunker. I could go on and on. Add into the mix that there are over 5,000 diagnostic tests that can be ordered to aid in getting to a correct diagnosis, and you begin to see the challenge with which us physicians are faced.

As medical students and residents, we were all taught to think in a certain way when trying to arrive at a diagnosis. An essential skill is learning to build a differential diagnosis. We doctors are, in general, pattern recognizers. Fever plus vomiting and diarrhea, plus stomach cramps usually equals a stomach virus. But, in addition to the most probable diagnosis, we think of other illnesses or conditions that could present in the same way, considering each one as a possibility. We then rule in or rule out, using further lab testing, or exam findings, or history gathering, until only one diagnosis is left.

I teach residents and medical students to build their differential diagnoses in one of two ways. The first way is to start with the most common, most likely disease at the top of the list, with other items in the differential diagnosis in descending order of likelihood. This is the most usual way of thinking. After all, common things are common; cough, shortness of breath and wheezing usually represents asthma.

There is another way to think, informally called "ruling out the badness." For a few presenting complaints, there is a possible diagnosis that is lethal, and even though it may not be common, it must be the first and most pressing diagnosis on the list. For example, if a teenager shows up with fever, chills, is ill-appearing and has petechiae, he could have life-threatening meningococcemia.

There are other, less worrisome illnesses that can show up in this way, but this is the dangerous, possibly lethal one, and it must be top of mind. All thoughts and actions are focused on proving if this is the diagnosis or not, with other things being secondary. My favorite professor from medical school called this stage "when the thin film of sweat forms on your brow."

So although our patients may not see it happening, every time a doctor sees a sick patient, he or she is mentally building a differential diagnosis, to include enough possibilities to ensure hitting the nail on the head and reaching the correct diagnosis. Initially, doctors must cast a wide net but still be able to separate the most likely or life-threatening diagnoses from the esoteric or least likely ones. Part of the job is to have enough illnesses on the differential diagnosis list, but to not get distracted by unlikely ones along the way.

Diagnosis is also a challenge because healthcare delivery is complex. Patients often transition from one place to another in the process of care (physician's office to the hospital, emergency department to the inpatient unit, inpatient unit to ICU), and in spite of best efforts, some details of information can be lost along the way. In the U.S., universal medical

records do not exist. Some important information may be known by one doctor, but not accessible to another specialist seeing the patient elsewhere.

Additionally, doctors are not foolproof; we make the same kind of unconscious mental errors everyone else makes in their everyday lives. Cognitive errors and biases affect everything we do. We sometimes jump to conclusions, subconsciously de-emphasize an important bit of information, or fail to think broadly enough, all with the best of intentions.

We also depend on our patient's ability to communicate, to answer our questions fully and thoughtfully, to listen to us, and to tell the truth (even if it is embarrassing). Increasingly, patients may speak another language, and then we use medical interpreters to help. This adds another layer between us and our patient, and sometimes things can be lost or garbled in translation.

All of us can recall a time we missed a diagnosis, or were slow or late in figuring out what was going on with our patient. We are generally plagued by these memories, since we seek (but never reach) perfection.

I have a clear memory of the young lady I took care of, a known asthmatic, who was admitted with an asthma exacerbation. Her mother mentioned to me that she had lost weight and no longer liked to eat. When I asked my patient directly, she silently held up her phone, where she had texted, "too tired to talk."

I thought I was dealing with another cranky teenager. I tucked her in, with usual hospital asthma treatment, and she was better the next morning. To further work up her weight loss and eating refusal, I ordered a barium swallow, thinking perhaps she had achalasia or a stricture. She aspirated the barium.

Further evaluation led to a diagnosis of myasthenia gravis. She had been having slowly progressive afternoon weakness, fatigue and difficulty in chewing and swallowing, which her mother had misinterpreted as laziness. And, it turned out, she wasn't really asthmatic; she was chronically aspirating due to muscle involvement.

I learned a lot with this case. First of all, labels are sticky, and the longer they are on the stickier they get. She had been labeled as an asthmatic, so other subtle signs were missed. Also, I misinterpreted her "too tired to talk" as sullen teen, not someone with had neuromuscular fatigue. Additionally, both her mother and I were worried about anorexia, since it is increasingly common in adolescent girls.

So we can never let our guard down. The next patient we see may the one who is not what they seem to be. This difficulty in diagnosing, this need to be part detective and to always be on our toes, this is part of the terror and the charm of practicing medicine. And getting it right - that's our greatest satisfaction! ■