My patient had been short of breath for a long time. For years, his asthmatic bronchitis had led to escalating doses of inhalers and more frequent courses of steroids. But now he seemed to be resistant to the tricks that had worked before. Regular albuterol, pulmicort and higher doses of prednisone weren’t rescuing him from his struggle to breathe. He walked into my office, pausing every 10 feet to sit on his walker and catch his breath.

Mentally, I scoured my therapeutic armamentarium and came up with nothing. Finally, grasping for the unlikely when the usual wasn’t working, I said, “I guess we can check a D-dimer to rule out the distant possibility of a blood clot to the lung.” The man had had a deep venous thrombosis but that was years ago, and he had no chest pain, no sudden increase in dyspnea. Despite his struggles, he was maintaining his oxygen level without supplemental oxygen. Later that day, the D-dimer returned elevated and a CT scan of the chest revealed multiple pulmonary emboli. Once again, my most dread diagnosis had almost thrown me and my patient a fatal curve ball. Luck, as much as diagnostic brilliance, had bailed us both out.

Every physician has diagnoses they dread. We are all trained to fear missing any diagnosis but the consequences of error frequently can be as minor as a slight delay in treatment or a sheepish grimace when a colleague makes the right call. For me, dreaded diagnoses are the ones where my misstep can kill a patient—the mundane neck pain subtly signaling the aortic dissection, the nausea and vomiting that turns out to be a myocardial infarction masquerading as gastroenteritis or the “migraine headache” that ends up being an intracranial hemorrhage. As diagnosticians, we tiptoe through minefields of lethal possibilities and hope we step in the right place.

Some would say their most dreaded diagnosis is no diagnosis. We all see patients with exhausting histories of confusing symptoms, who’ve had reams of tests and multiple doctor visits, who come to us looking for even just a label to explain their pain. Sometimes we strike pay dirt, finding the obscure “zebra,” such as some of the cases in this month’s case reports, that finally gives the sufferer at least an explanation for their symptoms and maybe also a treatment. Too often, we fail like diagnosticians before us and the patient trudges home with their misery and we trudge home wondering perhaps if we had read one more journal or textbook maybe …

Not making a diagnosis or making the wrong diagnosis is not only painful for doctor and patient, it can be an expensive provocation for legal action, which is why educational and insurance institutions are focusing on the diagnostic process, defining what works and what doesn’t, where doctors shine and how they fall short. Interestingly, one more journal article is rarely the explanation. Instead, aside from system errors, physician missteps result from disorders of attitude, taking shortcuts to conclusions, not following up on test results or being content with the obvious. If you don’t force yourself to think outside the box you’ll never see what’s outside the box.

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