Top Ten Hard to Diagnose Diseases

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Failure to diagnose and delay in diagnosis are some of the most common allegations of medical liability claims that we see at MagMutual. There are several known disease processes that are notoriously difficult to diagnose either because they mimic other, milder disease processes or their presentation leads the practitioner away from the correct diagnosis. Frequently this delay or misdiagnosis can lead to devastating consequences.

What is the patient safety and risk management point of view? What diseases do physicians struggle with diagnosing? Is there a common theme and why are they hard to pin down? Is there a means to diagnose these illnesses earlier?

1. **Pulmonary Embolus** - PE is associated with in-hospital mortality and it frequently occurs in post-op situations and after hospital discharges. There may be legitimate debate as to the extent of VTE prophylaxis required in a specific case. However, when adverse outcomes arise, your defense will be bolstered by your documentation showing that you considered, in a reasonable fashion, the risks and benefits of the level of prophylaxis you chose. Aggressive attention to ambulation and drug intervention reduces PEs by 60 percent. So even if you are doing everything to prevent these diseases, they may occur. And it may be a masquerader, often misdiagnosed as a myocardial infarction or pneumonia.

2. **Necrotizing fasciitis** - The “flesh-eating disease” is a rare infection of the skin and subcutaneous tissues, spreading along fascial planes. It progresses rapidly and has a greater risk of developing in the immunocompromised. It is of sudden onset and needs to be treated immediately with surgical debridement and
intravenous antibiotics. At the outset it can look like a more routine cellulitis. Major clues can be pain out of proportion to history and exam, and abnormal vital signs (toxic appearance) in the setting of musculoskeletal pain.

3. **Vascular pathology in the neck** - Compromise of cerebral perfusion secondary to disruption of blood flow in the neck can lead to devastating neurologic injury. There are a range of insults to the vasculature that can occur including clotting, embolism, trauma, and dissection. These manifest in various presentations. They can be silent, painful, or show vague neurologic symptoms. Investigation may require imaging that is not available in the necessary time at all institutions.

4. **Compromise of the spinal cord** - This includes epidural abscess and hemorrhage, spinal cord ischemia, and discitis. Any lesion around the spinal cord has a high risk of severe complications. Clinical presentations may be quite variable, and these rare conditions are mimicked by many common conditions. The clinical triad of fever, back or neck pain, and neurologic deficit may not be present in many patients with epidural abscesses or discitis, or may present when the time frame for intervention to prevent permanent deficits has passed. Early presentations will likely be subtle and atypical. A sequential evolution has been described, with localized spinal pain, radicular pain and paresthesia, muscular weakness, sensory loss, sphincter dysfunction, and finally paralysis. Abscesses from hematogenous spread tend to progress rapidly, while the signs and symptoms of osteomyelitis or discitis may evolve over weeks or months. Frequently, the patient gives a history of back strain or mild injury, further confusing the ability to make these rare and serious diagnoses.

5. **Ischemic bowel** - Intestinal ischemia is a condition in which inflammation and injury to the intestine results from inadequate blood supply. Intestinal ischemia can be either acute or chronic and occurs with greater frequency in the elderly. Intestinal ischemia spans a wide spectrum of severity; most patients are treated supportively and recover fully, while a minority with very severe ischemia may develop sepsis and become critically ill. Diagnostic tests including CT can be normal in early disease, making this a hard disease to diagnose. Abdominal pain out of proportion to abdominal exam should be a red flag to consider this diagnosis.

6. **Sleep apnea and post-op hypoventilation** - Accompanying the obesity epidemic is an epidemic of sleep hypoventilation syndrome. Sleep apnea is a sleep disorder characterized by pauses in breathing or instances of shallow or infrequent breathing during sleep. In obstructive sleep apnea (OSA), breathing is interrupted by a physical block to airflow despite respiratory effort, and snoring is common. Post op, these patients can face particular difficulties. Giving opioids or other respiratory depressants may alter their typical sleep pattern and lead to aspiration and hypoventilation events.

7. **Compartment syndrome** - Compartment syndrome is increased pressure within one of the body’s compartments which contains muscles and nerves, it most frequently occurs in the arms or legs. Trauma to an extremity can cause high pressure in a compartment, which results in insufficient blood supply to muscles and nerves. Acute compartment syndrome is a medical emergency that requires surgery to correct. If untreated, the lack of blood supply leads to permanent muscle and nerve damage and can result in the loss of function of the limb. Classically, there are “5 Ps” associated with compartment syndrome: 1) pain 2) paresthesia 3) pallor 4) pulselessness and 5) paralysis. Pain on passive range of motion is one of the first signs of compartment syndrome. A high index of suspicion is essential for timely diagnosis. Nerve blocks and high-dose narcotics desensitize the patient and may contribute to a delay in diagnosis. Loss of function and decreased pulses, however, are late signs. According to an article in Trauma[1], paresthesia in the distribution of the nerves transversing the affected compartment has also been described as a relatively early sign of compartment syndrome.

8. **Perforated or injured bowel post procedure** - Bowel injury may occur during surgery and is often occult. These injuries may be mechanical or thermal in nature. Often these are subtle injuries and are difficult to discern when the patient has post-op pain and gas issues. They may take a few days to manifest and become apparent. Delayed diagnosis can lead to serious adverse outcomes, including death.

9. **Appendicitis** - A common clinical illness that is a frequent miss in the ER or the office. There are many descriptions of classical signs or symptoms, but atypical presentations are common. Is the pain not at McBurney's point? Is it in the groin or higher in the abdomen? Does it mimic ovarian disease? Has the pain been lingering for several days, moving one away from that diagnosis? Is the patient obese, making the physical exam difficult? Clear discharge instructions and reexamination when the diagnosis is not clear can be helpful.

10. **Sepsis** - Sepsis affects more than 750,000 people annually, with a prevalence of three cases per 1,000 persons. Mortality rates remain between 25 to 30 percent for severe sepsis. Sepsis is responsible for 20 percent of all in-hospital deaths each year, which equals the number of annual deaths from acute myocardial
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infarction. Patients at the extremes of age are at higher risk of developing sepsis. Patients older than 65 years are 13 times more likely to develop sepsis and have a twofold higher risk of death from sepsis. Syndromes that mimic sepsis include hypovolemia, PE, acute myocardial infarction, acute pancreatitis, diabetic ketoacidosis, and adrenal insufficiency. Many patients present with a fever, and discerning those with sepsis is tricky. To diagnose sepsis, physicians must obtain historical, clinical, and laboratory findings indicative of infection and organ dysfunction.

What are the take-home lessons?

1. Some of these illnesses are common such as PE and appendicitis. In frequent illnesses, it is often the atypical presentations that are the problem. Subtle or unusual presentations can mislead you.
2. In rarer conditions, there is a narrow window of opportunity to make the diagnosis before it may cause irreversible harm to the patient. You might be diagnosing a disease that you have not seen before. The pearl here is to revisit your differential diagnosis in patients who are seriously ill and evolving, and try to avoid the anchoring bias that may lead you to not consider the real diagnosis.
3. In confusing situations, always ask yourself, “What else could this be? What would I hate to miss?”
4. What is the window of opportunity for having a realistic chance of altering the course of the care and preventing harm? The window of opportunity arises from a balance of the trajectory of the disease and the medical intervention capacity. Trajectory is defined as “at a given point in time, what is the likelihood of it leading to a medically or surgically irreversible harm or death?” Medical intervention capacity is defined as the “likelihood that medical or surgical intervention can make the diagnosis and subsequent intervention can change the course of the disease before leading to harm or death.” Many factors affect both the trajectory/likelihood of successful intervention and the medical intervention capacity to make a timely diagnosis and deliver the correct medical or surgical therapy. There can be instances in which there is practically no effective window of opportunity for a reasonable practitioner in a given setting, and the defense of such an outcome rests greatly on our ability to recreate (from the medical record documentation) what happened, what you thought, and your reasonable efforts.

[i] Trauma October 2006 vol. 8 no. 4 261-266

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