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# How to diagnose a rare disease when Occam's razor fails

Occam's razor, also known as parsimony, is the scientific and medical principle that the simplest explanation that addresses all the data or symptoms is usually the most likely diagnosis. The principle of parsimony is meant to ease diagnostics by leading clinicians to follow observable symptoms towards a single unifying diagnosis. As 13th century Franciscan friar and theologian William of Ockham [explained](#) when writing about his Aristotelian-based theory, "Plurality is not to be assumed without necessity [...] it is futile to do with many what can be done with few."

But does that line of reasoning always work? As Dr. James Kelly [writes](#) in "The Diagnostic Approach in Complex Patients: Parsimony or Plenitude?": "Atomic theory was initially rejected as it inferred the existence of invisible particles that had not yet been detected." With that in mind, what diagnoses have been missed due to a clinical preference for Occam's razor?

As healthcare workers, it's crucial to know when to use Occam's razor when diagnosing patients and when to pursue more complex leads based on your knowledge and intuition.

## **The appeal and limitations of Occam's razor**

Why have doctors so long relied on the principle of parsimony? The appeal of using Occam's razor in diagnostics is clear: a single, unifying cause that explains all symptoms in one fell swoop and results in their treatment is an appealing outcome. If that doesn't identify the underlying illness, using Occam's razor to test diagnoses can still help clinicians rule out the more obvious potential causes of a patients' symptoms.

However, despite the appeal of a single diagnosis, that is not always what patients are facing. What about cases with multiple comorbidities? Or when the disease being diagnosed is a rare condition? Or one where symptoms are less visible?

Complex and rare cases can present challenges when seen through the lens of parsimony, and seeking the simplest

explanation that covers all the symptoms in such situations may lead to serious misdiagnoses. Dr. Kelly [continues](#),

*“Diagnostic parsimony may be associated with under-ascertainment of secondary diagnoses in patients with chronic diseases. [A study evaluating] how often older patients with an established chronic disease were likely to be treated for a second unrelated prespecified diagnosis in comparison with matched patients without the chronic disease [...] showed that treatment for a second diagnosis occurred 30%-60% less often in the chronic disease group. It seems likely that under-diagnosis was a factor here. [...] The law of parsimony is gradually replaced by the law of plenitude as age, frailty, and comorbidities increase[.] Those involved in the care of patients with polymorbidity would do well to heed the words of Francis Crick, who observed that ‘Occam’s razor... can be a very dangerous implement in biology.’”*

### What happens when Occam’s razor fails?

Patients depend on clinicians’ accuracy and insight for their health, so stakes can be very high in diagnostic situations — literally life and death.

While working in an emergency department in Wollongong, Australia as a medical student, Fahad Farooq encountered what he was told was a simple case of a woman experiencing anxiety over a rash on her foot. He explained in [“A Close Cut: Occam’s Razor and Developing Intuition”](#) that the patient “muttered something quickly” about discomfort in her shoulders while Farooq inspected her swollen foot, the latter of which the attending nurse and resident focused on as the sole cause of her issues. Initially he agreed with his coworkers, opting to assume the correctness of Occam’s razor in this case.



When discussing the case and making his documentation soon after, however, doubt kicked in. Despite initially being hesitant to “add to [the] workload” of the resident, Farooq’s intuition pushed him to speak up since, he said,

*“Something just did not feel right. And if I did not say something now, I knew I would be up all night. I convinced myself to ask the busy nurse to obtain a bedside electrocardiogram.”*

Farooq’s persistence led to a revelation:

*“I was still a student and in no real position to be telling an experienced nurse what to do. But I trusted my instinct. Ten minutes later, I was looking at a non-ST-elevation myocardial infarction. The patient we had, in many ways, dismissed [as a case of anxiety] was having a heart attack in our consult room.”*

The impact of his intuition-driven decision to speak up and doubt the “simple” answer staggered Farooq. He explained,

*“The razor I had so necessarily trusted had dulled a little. It was the first moment, 4 years into medicine, that I felt my presence had directly impacted*

*someone’s life. Not just hers but my own. It put into perspective how suspicions for serious conditions must remain high, no matter the patient’s circumstances. I was forced to trust my patient and my gut, and doing so saved a life.”*

Though Farooq’s encounter with the limitations of Occam’s razor was in the context of a rather common condition — a heart attack — parsimony can also have limitations when it comes to rare diagnoses. This can be the case with both misdiagnosing a rare disorder in lieu of a more common one that would explain the symptoms or, in the opposite circumstance, attributing a common comorbidity to a pre-existing rare disorder.

The latter was the case with a two-year-old patient in Pakistan [described in Cureus](#). The toddler’s urinary tract infection was dismissed as being part of her pre-existing diagnosis of epidermolysis bullosa (EB), which can cause symptoms similar to a UTI. This case, as Dr. Abdur Rehman et al. explain, “emphasizes the importance of Hickam’s dictum,” the idea that patients can have multiple diagnoses (often referred to with the phrase, “as many [diagnoses] as they damn well please”),

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*“In the given case, the involved team initially bludgeoned the diagnostic possibilities into Occam’s razor. Said otherwise, it was easy to explain urinary symptoms under the umbrella of [EB], rather than considering it to be a separate unrelated entity. A patient can have multiple pathologies contributing to the same presentation. Clinicians, therefore, should keep an open mind regarding the possibility of coexisting illnesses, once the first diagnosis has been confirmed.”*



### **When Occam’s razor dulls, follow your intuition**

What is the solution to pursuing accurate diagnoses in light of this dulling of Occam’s razor? Keeping an open mind and following clinical intuition, say Drs. Nathan Borden and Derek Linklater in their article, [“Hickam’s Dictum.”](#) They describe a fifteen-year-old patient who was experiencing both appendicitis and pyelonephritis, a dual diagnosis that would be missed by following the principle of parsimony. They explain,

*“Physicians are reminded to pursue alternate or additional diagnoses when the patient’s symptoms cannot be explained by a single, unified diagnosis[.] Thinking broadly in the face of contradictory data helps to prevent the most common cognitive error in medicine: satisfaction of search. Satisfaction of search refers to the tendency to stop looking once something is found. Attributing the right lower quadrant pain to cystitis conforms to Ockham’s razor, but leaves one vulnerable to satisfaction of search, and in this case would have missed an additional diagnosis.”*

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*The sound of hoofbeats, as it is often said in medicine, can indicate a zebra or a horse.*

The importance of listening to patients, paying attention to their comments, and investigating invisible or less visible symptoms cannot be overstated. It is also crucial to learn when not to ascribe medical symptoms to a psychological cause, a common dismissal which can not only disenfranchise and even gaslight patients but can also often lead to a missed medical diagnosis — as was the case with the myocardial infarction patient mentioned earlier whose symptoms were repeatedly dismissed as being symptoms of a psychological anxiety disorder.

Sometimes it all comes down to a matter of listening to your gut and rejecting the simple hypothesis despite “satisfaction of search” — the sound of hoofbeats, as it is often said in medicine, can indicate a zebra or a horse.

### **Continuing medical education for diagnosing complex and rare disorders**

Establishing a [reliable system for diagnosing rare diseases](#) is crucial for the times when the hoofbeats suggest the presence of a zebra rather than a horse — or, to put it in literal terms, when the patient is experiencing a rare disorder rather than a common one.

Opening your mind to the presence of rare diagnoses is important, as is informing yourself about the different rare disorders relevant to your field. Here are several [continuing medical education courses](#) for when your patient is experiencing a rare disease or a complex series of symptoms with multiple causes:

1. [Differentiating Medical From Psychiatric Symptoms](#) — Learn to distinguish psychological and medical symptoms from one another
2. [Strategizing Complex Polypectomy](#) — Choose an appropriate resection technique based on polyp characteristics to improve management of complex polypectomy
3. [Complex Urologic Stones](#) — Improve the performance of minimally invasive renal surgery and robotic transurethral surgery