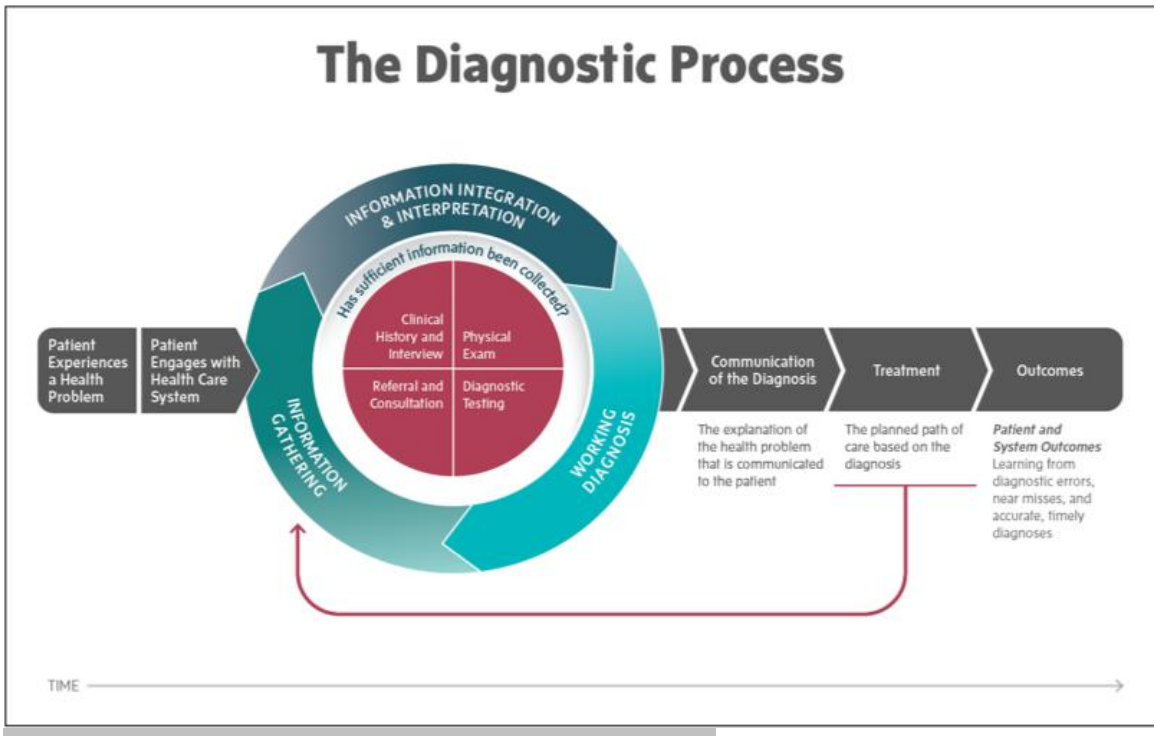
dear residents

Diagnostic Dilemmas **December 17, 2023**

Dear Residents,

Last week, I virtually attended a meeting of the **National Academy's Forum on Advancing Diagnostic Excellence**. We had a wide-ranging discussion and settled on a few high priority ideas to explore further. The diagnostic process involves every aspect of the healthcare system.



The National Academy of Science Engineering and Medicine

The path to diagnosis in medicine has substantially deviated from the traditional sequence of history->physical examination->clinical reasoning->selection of tests->working diagnosis. More often than not, we now start with lab results and imaging, and in doing so, we work backwards from a differential diagnosis of the particular lab or imaging finding, rather than work forward from a list of possible diagnoses derived from the patient's history and physical examination. How does this happen? In emergency rooms across the country, the chief complaint may lead to protocol-driven (and ostensibly efficient) bundled order sets without the prior application of clinical reasoning. Syncope = head CT. Dyspnea = CT angiogram. Chest pain = troponin. Some of this is driven by the minimax principle – minimize the maximum possible loss - envisaging the worst-case scenario even if that scenario is rare.

This upends the diagnostic process grounded in ideas such as pre-test probability, post-test probability, likelihood ratios, sensitivity, specificity, positive and negative predictive values. It is being replaced with the need to work backwards from a positive result or imaging finding. Imagine if your list of patients for the day were listed by their lab or imaging finding rather than by the clinical reason for the visit:

1. Positive ANA
2. Renal cyst
3. High CRP
4. L2-L3 Interverbal disc herniation
5. Gallstones
6. High CEA
7. Positive Lyme antibodies
8. Positive urine culture
9. Negative cardiac stress test
10. High creatine kinase

Reverse interpretation of such findings is now our daily work, and we are **compelled to reconstruct the significance of the results**. We find ourselves going down the rabbit-hole of a testing and imaging adventure. I made up some hypotheticals for your consideration. Some are straightforward, others will require additional thought. The point is that the test alone (with some rare exceptions) is insufficient to make a diagnosis. Additionally, it is hard to stop working up positive test results even when they are of low specificity or of unclear significance.

1. Positive ANA – a 70-year-old male with mechanical right knee pain.
2. Renal cyst – CT finding in 45-year-old woman imaged after a motor vehicle crash.
3. High CRP – 63-year-old male with coronary artery disease.
4. L2-L3 interverbal disc herniation – MRI finding in a 32-year-old swimmer with low back pain.
5. Gallstones – US finding in a 51-year-old woman with dyspepsia.
6. High CEA – 44-year-old male with a positive FOBT.
7. Positive Lyme antibodies – 39-year-old male with fatigue.
8. Positive urine culture – 35-year-old woman with menorrhagia.
9. Negative cardiac stress test – 55-year-old male with new onset angina.
10. High creatine kinase – 22-year-old black male with a tennis elbow.

In addition to the overuse of diagnostic tests, there remains the problem of their underuse in certain populations leading to **diagnostic inequity**. Diagnoses have i**mportant implications** for insurability, reimbursement, and the provision of resources. Diagnoses are also labels that have **societal implications**. We may be forced to confer diagnoses (or commit to them) to navigate the rules of the medical-industrial complex or withhold them to prevent harm or stigma. If I wanted to treat a patient with undifferentiated something something with this or that medication – I may need to commit to a diagnosis required to prescribe that medication. If your patient is in a profession that would cause them to lose their job with a particular diagnostic label, you might want to be certain before you did so – seizures, depression, cognitive impairment, etc.

Some diagnoses are temporary mental workstations that facilitate further clinical reasoning or support the gathering of resources. I learned early on to remain diagnostically cautious. Rather than “pneumonia” the more strategic diagnosis may be “right lower lobe infiltrate” – such an approach prevents premature diagnostic closure. There are many comparable examples such as “UTI” versus “pyuria.”

Diagnostic error remains one of the more important causes of patient harm – failing to make a diagnosis or making an erroneous one has myriad consequences, from missed malignant lung nodules to socially charged diagnoses like factitious disorder. Making and assigning diagnoses to complex, rare and contested conditions remains a challenge and some of our “thinking workstations” acquire the trappings of a firm diagnosis – all those “not otherwise specified” (NOS) diagnoses which then threaten to become an end in themselves rather than as waystations for further inquiry. I remember Dr. Herb Fred erupting with disdain at “diagnoses” we threw around in morning report such as “transaminitis” or “failure to thrive.” He felt that this was terribly lazy of us. Some NOS diagnoses are very useful like

“fever of unknown origin” while others are simply artefacts of our flawed disease classification systems or just feel too convenient to not use.

Artificial intelligence holds promise in decreasing missed diagnosis, or in discovering rare diseases, but the algorithms themselves may have embedded errors and inequities – they may inadvertently perpetuate such problems. If we can detect and mitigate systemic error, combined with advances in data science, we may find our work greatly augmented by technology Perhaps AI will be better at diagnosis than us and perhaps chatbots will have better communication skills than we do. I recognize that we are in a changing world and that it sometimes gets hard or even impractical to follow traditional diagnostic pathways, but I do believe that we will not outgrow the value of patient first, tests second.

Warm regards,

Dino Kazi