Diagnostic reasoning: a single entity diagnosis is often inadequate

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I enjoyed Brush and colleagues' review of diagnostic reasoning, especially the figures illustrating the relationship between pre-test and post-test probabilities.[1] An educational review should, however, emphasise that a diagnosis is often not a unitary concept such as "aortic dissection", even in cardiology.

This is particularly evident in paediatric neurology, where it is important to not just make a diagnosis of the single most likely condition, but to formulate a differential diagnosis as well- for example, when rapidly diagnosing Guillain-Barré syndrome keep the important differentials of acute myelopathy and even botulism in mind, Very occasionally, in a long career, these less likely differentials will prove to be correct.

Additionally, diagnoses can be nested, with a spectrum of increasingly specific diagnoses emerging as more clinical, imaging, and laboratory data are accrued. An infant in respiratory distress in the emergency department, can be quickly diagnosed with a congenital lactic acidosis, then later with Leigh's syndrome, then even later with complex 5 deficiency: a respiratory chain disorder, then later still with a mitochondrial disorder because of a mutation in MT-ATP synthase F0 subunit 6, at position 8993. When discussing the revised more precise diagnosis and bleak prognosis you might be asked if the infant had a T to G or a T to C mutation. Such a distinction would be important to physicians only if it affected the management, prognosis, or genetic counselling. To some families, however, the precise identity of the disease can feel very important.

Finally, in paediatric neurology as in child and adolescent psychiatry a single entity diagnosis is often inadequate as several key processes can be contributing to the presentation at the same time. In these circumstances a multiaxial diagnostic framework or narrative formulation is needed to best inform management and prognosis.

Reference

1. Bush JE, JrSherbino J, Norman GR. Diagnostic reasoning in cardiovascular medicine. BMJ2022;376:e064389. Doi: 10.1136/bmj-2021-064389 pmid: 34987062.

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