Handout for Diagnostic Dilemmas

Diagnostic dilemmas occur daily in clinical practice. Some are straightforward and others are more problematic.

Things to consider in making a diagnosis

1. Know the specificity, sensitivity and positive and negative predictive value in evaluating labs.
2. Know that there is diagnostic error in radiologist reports
3. Putting pieces of the puzzle together
4. Always starts with a meticulous history and physical exam
5. Diagnostics need to ordered based on the history and physical.
6. Today, there is an increasing reliance of labs, with less reliance of the basics of health care
7. Remember, diseases do not read text books

Ordering lab tests

1. Why should they be ordered
   a. Confirmation to make a diagnosis
   b. Screening test if there is high prevalence of a condition in a certain population
2. Why should you not order them
   a. Parent wants testing done
   b. You are not clear what is wrong so you order a wide battery of tests
3. What will the lab test do to help you care for the patient?
4. What is the expense of the test?
5. Would I still order it if the patient did not have insurance?
6. Can I interpret the test?
7. Which is the best test to eliminate the differentials and determine the diagnosis?
8. What is the sensitivity and specificity of the test?
   a. The normal reference range is the mean ± 2 S.D.
   b. There is a 1 in 20 chance that the result is out of the range
   c. You need to understand the pitfalls of the test in order to interpret it and take the next step
9. Is it an error prone test?

Considerations about diagnostic testing

1. Clinical follow-up
   a. Great diagnostic test
2. If you must order a test, you need to understand:
   a. Laboratory reliability
   b. Must have the right person to interpret the test
   c. Must use an age appropriate reference
   d. The test must be helpful in the diagnostic process
   e. Understand what the lab looks like when it is highly suggestive of a disease
3. Sensitivity and Specificity, Positive and Negative Predictive Value
   a. The sensitivity of the test reflects the probability that the screening test will be positive among those who are diseased.
   b. In contrast, the specificity of the test reflects the probability that the screening test will be negative among those who, in fact, do not have the disease.
   c. As the disease becomes more prevalent, subjects are more frequently in the "affected" or "diseased" column, so the probability of disease among subjects with positive tests will be higher.

Case 1 points out the problems that can arise with the lab test you order and understanding this concept.

- Current vitamin D assays measures total serum 25-(OH)D
  - IOM recommends a threshold of 50nmol/L for bone health
  - International Osteoporosis Foundation and Endocrine Society recommends a threshold of 75 nmol/L
  - Deficiency associated with increased risk of chronic disease—CV, some cancers, autoimmune disease and infectious disease

- Different testing methods
  - Competitive protein binding
  - Lumipulse G 25-OH vitamin D
  - Immunoassay
  - High-performance liquid chromatography
  - Combined high-performance liquid chromatography and mass spectrometry (LC-MS/MS).

- Vitamin D
  - Sensitivity and specificity of these tests are unknown because of the lack of studies that use an internationally recognized reference standard.
  - Variability between assay methods and between laboratories using the same methods may range from 10% to 20%
  - Classification of samples as “deficient” or “nondeficient” may vary by 4% to 32%, depending on which assay is used.
  - 25-(OH)D may act as a negative acute-phase reactant and its levels may decrease in response to inflammation. (USPHSTF, 2014)

<table>
<thead>
<tr>
<th>Test</th>
<th>Sensitivity (true positives)</th>
<th>Specificity</th>
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<tbody>
<tr>
<td>Fujirebio</td>
<td>100% (95% CI: 71.3%-100%)</td>
<td>91% (CI: 83.1%-06%)</td>
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<tr>
<td>Abbott Assay</td>
<td>90.90% (95% CI:58.7%-98.7%)</td>
<td>97.8% (95% CI: 92.1%-99.7%)</td>
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Beckman Method  |  90.9% (95% CI: 58.7%-98.5%)  |  86.7% (95% CI: 70%-93.7%)  
Roche Method  |  72.7% (95% CI: 39.1%-93.7%)  |  94.4% (95% CI: 87.4%-98.1%)  

1. Making the diagnosis
   1. Could this be an unusual presentation of a common disease?
   2. Could this be a rare disease?
   3. Does my patient have more than one disease
      1. Where you hear hoof beats, think horses, not zebras
         a. You cannot neglect the possible zebra
   2. Perpetuating the diagnosis over time
      1. Confirmation Bias
         a. Clinician only seeks out information that affirms the diagnosis and excludes facts that contradicts the initial diagnosis
         b. If an expert feels this way, they may close out other members of the team
      2. Authoritarian Bias
   3. Affective Errors
      a. Emotions interfere with diagnostic judgement
   4. Attribution Error
      a. Minimize signs, symptoms, or lab data
      b. Use of slight before a description of something, i.e. Slight tachycardia
   5. Tendency toward benign thinking (Kliegman, Bordini, Basel, Nocton, 2017)

3. Overreliance on published criteria
   1. Kawasaki Disease
      a. The criteria was available before they realized that patient could have an atypical Kawasaki and with strict reliance on only the criteria, cases were missed and coronary artery aneurysm developed.
   2. Jones Criteria
      a. Were recently revised to stress the migratory, transient and disproportionately painful polyarthritis
   3. Marfan syndrome
      a. Mutation in the FBN1 gene has a wide phenotypic variation
      b. Only 12% of who do not meet clinical criteria have the mutation, but 66^ of those who have FBN1 mutation meet the clinical criteria

4. Developing the diagnosis
   1. Need to think broadly
   2. Think by system
   3. What is the most likely?
   4. Develop plan that will be the most helpful toward the diagnosis

5. Incidence of diagnostic error
   1. Elstein
      a. Diagnosis is wrong 10-15% of the time
2. Autopsy studies  
   a. 10%-20% had major diagnostic discrepancies
3. Second reviews  
   a. 10-30% of breast cancers are missed on mammography
4. Diagnostic testing errors  
   a. Error related to laboratory testing are the most common
5. Malpractice Claims  
   a. Problems related to diagnostic error #1 reason for malpractice
6. Types of Errors  
   1. Systems Error  
      a. Complex, inter-related pieces of the health care system  
      b. Communication breakdown  
      c. Care Coordination breakdowns  
   2. Cognitive errors  
      a. Problems with the clinical reasoning process  
         i. Knowledge deficit  
         ii. Failing to acquire to use the relevant data or put together the pieces
7. Analyzing the data  
   1. Abductive approaches  
      a. Look at positive and negative predictive values of various laboratory tests  
      b. Look at whether the diagnosis is less or more likely based on the results—Looks at sensitivity and specificity  
   2. Inductive approaches  
      a. Uses observations and works backward using pathophysiology  
      b. Testing strategy is developed by confirming or rejecting hypothesis  
   3. Deductive approach  
      a. Begins with general principle and aligns the patient’s presentation with the general principles  
      b. Use of disease based criteria
8. Improving Clinical Reasoning  
   1. Need for  
      a. Intervention to improve knowledge and experience, with simulation-based training, improved feedback and education  
         i. Improve expertise and experience  
         ii. Focus on specific content areas that you are working in  
         iii. Continuing education  
         iv. Routinely test competency in diagnostic accuracy  
         v. Find simulation exercise to increase your exposure to a variety of case presentation—journal, websites  
         vi. Get feedback from mentors  
         vii. Learn from mistakes  
      b. Intervention to improve clinical reasoning and decision making skills  
         i. Reflective practice
ii. Training in intuitive processing  
iii. Using a checklist  
iv. Train on typical pitfalls of specific clinical conditions  
v. SNAPPS:  
   1. Summarize history and findings  
   2. Narrow the differential  
   3. Analyze the differential  
   4. Probe preceptor about uncertainties  
   5. Plan management  
   6. Select case-related issues for self-study

c. Intervention to provide cognitive help including the use of HER with integrated decision support (Graber, et al., 2012)  
   i. Second Opinions-Get help from others  
   ii. Use guidelines, clinical algorithms, mnemonics to reduce reliance on member  
   iii. Improve the medical record  
   iv. Integrated diagnostic decision support

9. Role of patient  
   1. Take a more proactive role in their own care and safety to the extent feasible  
   2. Contributing to the healthcare system—give feedback about things that went wrong  
   3. Play a role in informing policy and research agendas to prevent medical errors (McDonald, Bryce, Graber, 2013)

Websites of interest

**Stimulation/real time websites**

- [http://www.med-u.org/](http://www.med-u.org/)
- NEJM video cases
- Reel Dx
- Human Diagnosis Project
- IMreasoning
- Figure 1

**Diagnostic decision Support Websites**

- Diagnosis pro
- Dxplain: [http://www.mghlcs.org/projects/dxplain](http://www.mghlcs.org/projects/dxplain)
- PEPCID:
- Visual Dx [www.visualdx.com](http://www.visualdx.com)

- Provider-focused
• Diagnose
• ID Dx
• Mobile DDx
• DSM-5 Differential diagnosis
• Diagnosaurus
• Patient-focused
  • Your RapidDiagnosis: Childhood skin rashes

Reduce your Diagnostic Error in Ambulatory Settings
• Make sure patients get the screenings they need
• Make sure all old records, tests result and consults are available
• Track all requested diagnostic tests and consults to make sure they are complete
• Have a great system to review all tests
• Follow up on patients that are recently hospitalized, seen in ED or in office for a new complaint
• Give patients copies of their results
• Use EMR and make sure the problem list is accurate
• Designate a surrogate to review returning test results for planned absences

Reduce your Diagnostic Error As NP
• Be reflective. Diagnostic time out
• Listen
• Learn causes of diagnostic error and how to avoid pitfalls
• Construct a differential diagnosis, do not always rely on intuition
• Make sure you partner with your patient
• Make sure you follow up on diagnostic tests and consults
• Speak with consults, pathologists, radiologist if you are not sure what is the best test to order
• Empower your colleague to let you know if they become aware of diagnosis you made that has changed. Know your diagnostic errors
• Get second opinions
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