Differential Diagnosis: Approaches and Pitfalls

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Learning Objectives

- Explore the theory and practice of differential diagnosis

- Understand the importance of using a systematic approach

- Learn how cognitive biases can lead to diagnostic errors

- Practice differential diagnosis skills using simulated pediatric cases

The Mystery

Chief complaint → ???? → Differential Diagnosis (Structured, Ordered)
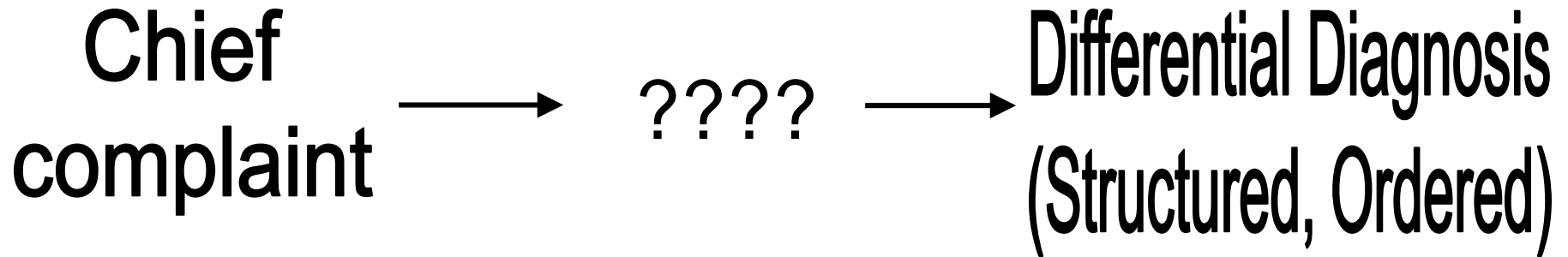
An Error-Prone Endeavor

"Most [medical] errors are mistakes in thinking."

   -- Jerome Groopman, M.D.

From Groopman, J. How Doctors Think. Houghton Mifflin Press, New York. 2007.

What is a Differential?

- An evolving list of diagnostic considerations
- But also a <u>rational and systematic approach</u> to the problem of evaluating and treating patients whose underlying disease is unknown

Differential Diagnosis: Goals

- Guide evaluation and treatment
- Generate working diagnoses
- Rule out life-threatening/time-critical conditions
- Make the correct diagnosis

Differential Diagnosis is Hard

- No one-size-fits-all approach to differential exists
- A "good" differential is not clearly defined (e.g., what to include/how to structure)
- Thousands of diseases to remember/consider
- Medical literature constantly changing

… And More Challenges

- Patients don't always provide full information (or we don't ask)

- Competing demands on time (we're busy)

- Lack of feedback/definitive confirmation of diagnoses

- Human reasoning inherently subject to error

Differential Process Research

- Two general approaches identified:
  - Pattern Recognition (Intuitive)
  - Analytic (Hypothesis-Driven)
- Approaches overlap and combinations of both are used in practice ("dual processing")
- Both approaches subject to error from cognitive biases

Pattern Recognition (Intuitive) Approach

- Match your overall patient assessment to your internal data bank of how diseases present
- "This looks like a patient with pneumonia…"
- Can happen rapidly/almost unconsciously

Illness Scripts

- The "story" of various ways diseases present
- Includes risk factors, signs and symptoms, time course, response to interventions, etc.
- Our illness script "data bank" gets richer from seeing more patients
- Part of why we tend to get better at differential with experience

Analytic Approach (Hypothesis Driven)

Collect Information

Generate/Test Hypotheses (Differential)

Analyze and Synthesize Data
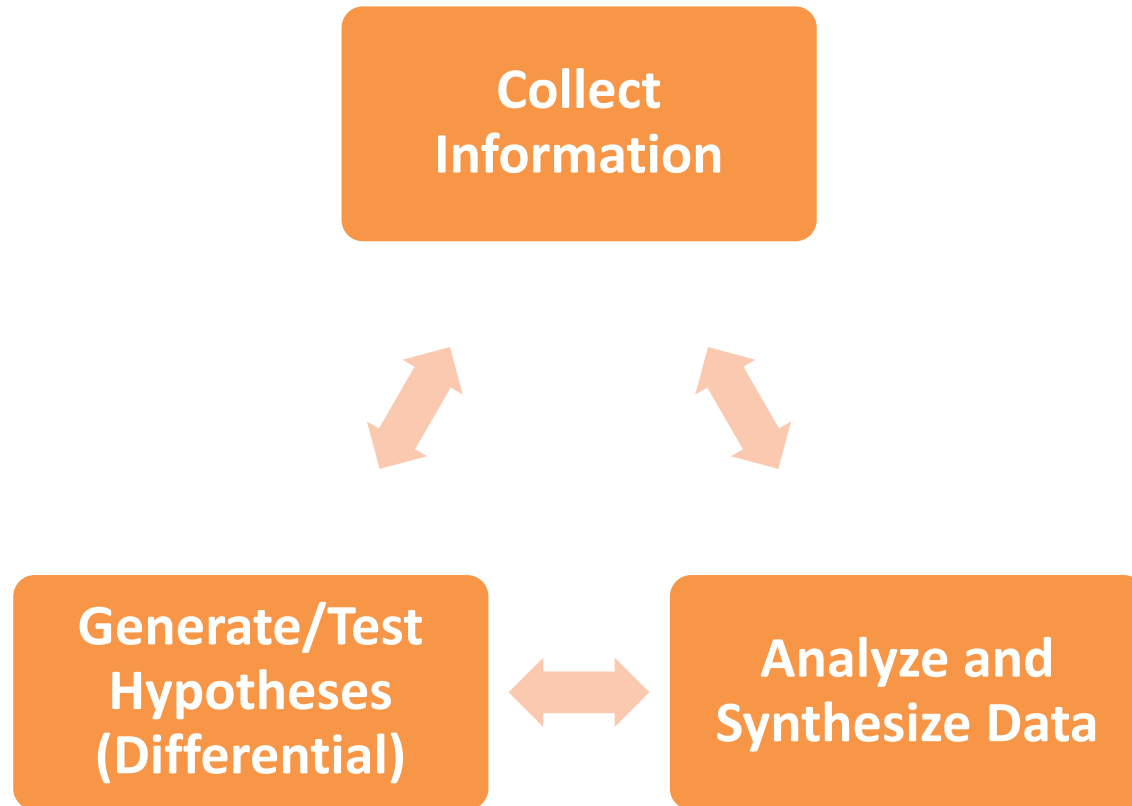
Hypothesis generation can be based on intuition, probability, pathophysiology, etc.

Analytic Process: Information Processing

Less

Information Processing

More

Raw Data

Key Features/Problem List

Clinical Syndromes

Diseases
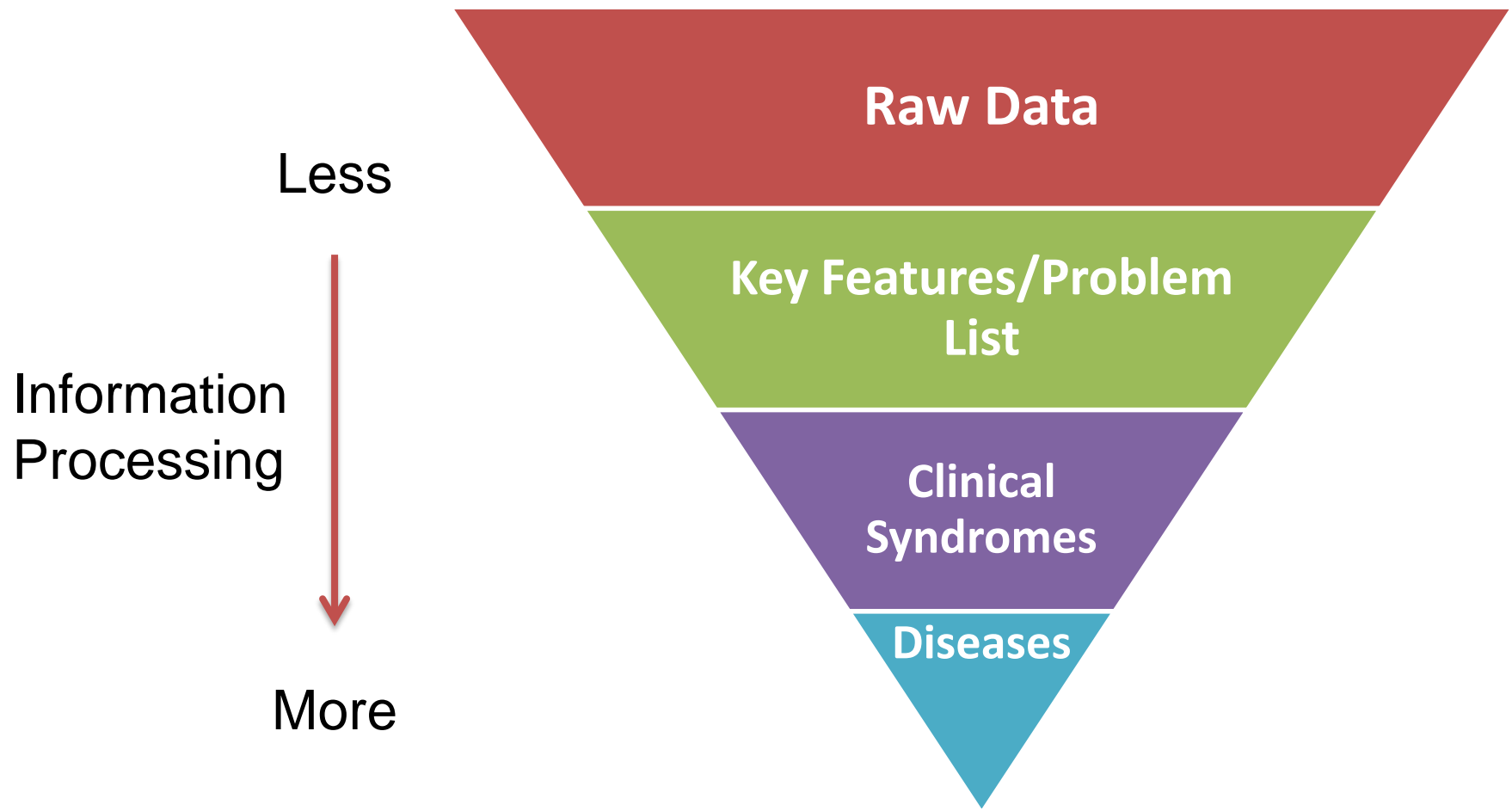
Cognitive Biases

- Recognized situational tendencies to make inaccurate judgments or decisions
- Mismatch between our perceptions or interpretations and objective reality
- Shortcuts in thinking/analysis make us particularly vulnerable
- Many biases identified in the literature

Cognitive Biases in Practice

Cognitive Bias	Example
Anchoring bias	Giving excess weight to early/initial information – e.g. accepting diagnosis handed off
Confirmation bias	Focusing on supporting evidence for diagnosis while ignoring contradictory evidence
Availability bias	Giving too much weight to diagnoses that come readily to mind, e.g. • Focusing on a diagnosis because that's what you've been seeing a lot of • Focusing on a diagnosis because you just heard of an interesting case of it

These are just a few examples of cognitive biases.

Differential Process Toolkit

1. <u>Identify</u> and <u>qualify</u> key features

2. Create a summary statement

3. Generate potential diagnoses <u>systematically</u>

4. Use references to help

5. Structure, order, and prune your list

1. Identify Key Features

- Part of the "art" of medicine.
- Some guides to consider:
  - Chief complaint (why?)
  - Defining features: Unusual features that point to defined set of illnesses (best case pathognomonic)
  - Discriminating features: Help rule disease categories in or out (e.g., well- vs. ill-appearing)
  - Don't overlook pertinent negatives

Qualify Key Features

- Turn the raw data into medically meaningful information

- Good qualifiers help further define distinct disease categories (clinical syndromes), e.g.

  "Fever since Wednesday afternoon." -> <u>acute</u> fever (vs. persistent or intermittent fever)

  "Vomiting everything. It looks green." -> (<u>bilious</u> emesis (vs. nonbilious emesis)

2. Create a Summary Statement

- Summarize the qualified key features to create a "problem representation"
- Goals are to
  - Frame key features
  - Eliminate extraneous information
  - Set the stage for the reasoning that follows

Example 1

- 18 month old boy with a rash and knee pain. The rash started 2 weeks ago as a small red area on his abdomen; now it is a larger red oval. Yesterday he started to complain about his knee hurting. Mom pulled a tick off him 1 month ago.

Example 1: Key Features

- 18 month old boy with a <u>rash</u> and <u>knee pain</u>. The rash started <u>2 weeks ago</u> as a <u>small red</u> area on his abdomen; now it is a <u>larger</u> red oval. <u>Yesterday</u> he started to complain about his <u>knee hurting</u>. Mom pulled a <u>tick off him 1 month ago</u>.

Example 1: Summary Statement

- Toddler with tick bite 1 month ago, now with <u>expanding</u> red rash and <u>acute</u> knee pain

Example 2

- Two year old boy with fever for eight days. He has cried a lot more than normal and is often difficult to console. Two days ago, it looked like he had pink eye, but there was no discharge. Today his hands look swollen. On exam, a 2 cm cervical node is felt on the left.

Example 2: Key Features

- Two year old boy with <u>fever for eight days</u>. He has cried a lot more than normal and is often <u>difficult to console</u>. Two days ago, it looked like he had <u>pink eye</u>, but there was <u>no discharge</u>. Today his <u>hands look swollen</u>. On exam, a <u>2 cm cervical node</u> is felt on the left.

Example 2: Summary Statement

- 2 year old with <u>persistent</u> fever, fussiness, <u>nonpurulent</u> conjunctivitis, large <u>unilateral</u> lymph node, and <u>acute</u> hand swelling

3. Generate Diagnoses Systematically

- For any patient, there are many potential diagnoses

- Systematic approaches help reduce the risk of missing important considerations

Big point: You will never make a diagnosis that you do not explicitly consider.

Example: Thinking Systematically

- Example: 3 y.o. with acute respiratory distress

- What is your differential?

- Where to start? We will return to this question shortly.

Situation-Specific Mnemonics

- Anion-Gap Metabolic Acidosis: MUDPILES
  - Methanol
  - Uremia
  - DKA
  - Paraldehyde
  - Iron/INH/Inhalants/Ibuprofen
  - Lactic acidosis
  - Ethanol/ethylene glycol
  - Salicylates

Mnemonics for Specific Situations

- Altered Mental Status: AEIOU TIPS
  - Alcohol
  - Endocrine/Electrolytes (including glucose)/Encephalopathy (hepatic)
  - Ischemic/Infarction/Intussusception
  - Oxygen (hypoxemia; also CO_2 retention)
  - Uremia
  - Trauma, tumor, temperature (high/low), toxin (drug)
  - Infection
  - Psychiatric
  - Seizure/Syncope

General System: VINDICATE

- Pathophysiology-based approach particularly well suited for new/challenging situations

  V – vascular
  I – infectious/inflammatory
  N – neoplastic
  D – degenerative/drugs
  I – idiopathic
  C - congenital
  A – autoimmune
  T – trauma
  E – endocrine

Other Causation-Based Systems

- Consider ways a specific finding can occur
- Example: Approach to a patient with low albumin
  - Not making enough (liver)
  - Not absorbing protein from gut
  - Losing in stool
  - Losing in urine

Lumping vs. Splitting

- Often helpful to start with broad categories (lump) then split into subparts, e.g.
  - FUO can have Rheumatologic vs. Infectious vs. Oncologic causes
  - Within infections, causes may be further divided into focal vs. systemic infections
- Split to the extent clinically helpful for evaluation and management

Example: Thinking Systematically

- Example: 3 y.o. with acute respiratory distress
- Try using VINDICATE to generate potential ideas

By using a systematic approach, you are likely to consider possibilities that otherwise may be missed

4. Supplement With References

- Whatever situation you are facing, references can help

- Most currently require you to identify 1-2 key features to focus your search

Where to Look Things Up

- Books – e.g. "Signs and Symptoms in …"
- Up-to-Date® (e.g., "Approach to the child with …")
- Cheap and quick: Google "[finding or findings] differential diagnosis"
- Other electronic resources: MD Consult, Skyscape (for PDAs/smart phones),
- The list of potential sources of information is nearly endless
- Any suggestions?

5. Structure, Order, and Prune

- Goal is a limited set of diagnostic possibilities (including at least one working diagnosis)
- Typically requires eliminating many considerations … or putting them on the back burner
- Need to assess the likelihood and urgency of different diseases

Time-Tested Pearls…

- Often said that "common things are common"
- Thus all else equal, you are more likely to see:
    1. Common Presentations of Common Diseases >
    2. Uncommon Presentations of Common Diseases >
    3. Common Presentations of Uncommon Diseases >
    4. Uncommon Presentations of Uncommon Diseases (this last case will be very difficult to diagnose!)
- Point: Don't overlook uncommon or incomplete presentations of common diseases

Occam's Razor vs. Hickam's Dictum

- Occam: All else equal, simpler explanations for the facts at hand are preferable to complex explanations
- Serves as a reminder to search for unifying diagnoses for the clinical presentation
- Does <u>not</u> imply that a single disease process will always explain everything
- Hickam: "Patients can have as many diseases as they [darn] well please."

Organize Your List

- Goals are to convey your
  - Logic (thought process)
  - Disease considerations ("differential")
  - Working diagnosis
- Highlight life-threatening diagnoses you wouldn't want to miss
- One strategy – present disease categories from most to least likely, and then specific relevant diseases within each category

A Final Check

- Has your clinical decision making been adversely affected by cognitive biases?
- Think about how your reached your conclusions
- Reduce your error risk by taking a diagnostic "time out"

The Diagnostic "Time-Out"

- How did I arrive at this diagnosis?

- Did I consider all the alternatives?

- Did I settle too early on a diagnosis?

- Did I accept someone else's diagnosis without question?

- And for admitted patients: does the diagnosis still fit the clinical picture?

Small Group Breakout

1. Identify and qualify the key features of the case
2. Create a summary statement that highlights key features
3. Generate and refine a differential and a working diagnosis.
4. Organize your differential presentation in a manner that reflects and conveys your thought process.

Final Thoughts

- Differential diagnosis is a <u>skill</u> that can be learned and developed
- You can only make a diagnosis that you explicitly consider
- Using systematic processes will make you less prone to error
- Time spent examining how you arrived at your differential is often well spent
- GO FORTH AND DIAGNOSE WELL!!

Case 1 – Example Summary Statement

- 10 year old ill-appearing girl with no significant PMH presenting with acute onset periumbilical abdominal pain, nonbilious emesis, fever, anorexia, and peritoneal signs.

Case 2 – Example Summary Statement

- 5 year old non-toxic appearing boy with no significant PMH presenting with acute periumbilical abdominal pain, monoarticular arthritis, and lower extremity palpable purpura several weeks after a URI

Case 3 – Example Summary Statement

- 1 year old ill-appearing boy with no significant PMH presenting with acute paroxysmal abdominal pain, lethargy, nonbilious emesis, decreased stool, and a tender, distended abdomen shortly after eating eggs for the first time

Case 4 – Example Summary Statement

- 3 yo non-toxic appearing girl who attends day care with acute vomiting followed by non-bloody diarrhea, stable vital signs, and a reassuring abdominal exam.

Case 5 – Example Summary Statement

- 16 yo female with a several month history of weight loss, fatigue, fever, headaches, and frequent ibuprofen use, now presenting with a second episode of acute abdominal pain, bloody stool, pallor, and dehydration

Case 6 – Example Summary Statement

- 9 year old boy with history of JIA, obesity, and steroid use, presenting with acute severe periumbilical abdominal pain, bilious emesis, anorexia, dehydration, and possible mild trauma