When you hear hoofbeats, think horses.... Unless a Zebra walks into your office

Presented by:
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• I have nothing to disclose
Objectives of this presentation

• Specify the incidence of rare diseases in primary and urgent care practices.
• Identify “red flags” and atypical outcomes when evaluating what appears to be a benign condition
• Audience participation sharing of PEARLS of wisdom
• Describe the role Nurse Practitioners and other health care providers have in managing zebras in the primary care setting.
What is NOT going to happen in this lecture

- Extensive pathophysiology for each condition listed
- Treatment protocols for each rare illness
- A ZEBRA CRYSTAL BALL door prize
Diagnosis

Dr. Lisa Sanders recreates hard-to-solve medical case studies.

Latest

Aug 29, 2018

She Had a Long and Strange Rash on Her Arm. What Was It?
It didn't hurt, but the woman wasn't sure whether the streak on her arm was harmless or potentially dangerous.
By LISA SANDERS, M.D.

Aug 15, 2018

Her Searing Gut Pain Suggested Celiac. Why Didn't Cutting Out Gluten Help?
The patient, a retired nurse, had no insurance and diagnosed herself — which may have been part of the problem.
By LISA SANDERS, M.D.

Aug 10, 2018

A Diagnosis Update: A Young Woman's Extreme Muscle Pain Persists
More than 2,000 readers suggested diagnoses for Angel, the 23-year-old who continues to experience muscle pain and darkened urine.
By LISA SANDERS, M.D.
A Lisa Sanders case walks in the office:
27 yr. old Caucasian male

- Previous provider:
  - Labs (WNL)
  - Acupuncture
  - No other workup

- Here for a 2\textsuperscript{nd} opinion
  - Weight loss 100 pounds
  - Nausea/Vomiting
  - Fatigue

- PMH:
  - anxiety/depression, vitiligo
- PSH: none
- Family hx: none
- Medications: Zofran, marijuana for anxiety
Mystery man…

• What are some of your thoughts before you go into the room?

• More questions?
• Diagnosis ideas?
• Vital Signs?
More information

- Previous weight: 285, now 175 lbs.
- HR 98
- Temp: 98.7
- Blood pressure: 98/55
- O2: 98
Addison’s Disease

- Adrenal gland dysfunction - hypocortisolism
- 1 in 10,000 persons
- Fatigue, hypotension, craving for salt
- Hyperpigmentation
- Recognizing ADRENAL CRISIS

- Labs:
  - Hypoglycemia
  - Hyponatremia
  - Hyperkalemia
  - Hypercalcemia
  - Cortisol testing?
  - Not practical in urgent care setting
What is a rare disease?

- “Any disease, disorder, illness or condition affecting fewer than 200,000 people in the United States is considered rare by the National Institutes of Health (NIH), the U.S. Food and Drug Administration (FDA) and NORD.” Source: NORD, 2018

- 7000 rare diseases in the US.
- 30 million Americans are affected or roughly 9% of the population.
- 9.4% of the population has Type 2 Diabetes
So how often will I see a rare disease?

- 7000 rare diseases in the US.
- 30 million Americans are affected or roughly 9% of the population.
- 9.4% of the population has Type 2 Diabetes.
- One person in 1250
- RARE DISEASES ARE COMMON
- They will happen and you will never forget them.
Medicine is often taught as heuristics
RULES OF THUMB

• Pattern Recognition
  – Woman with back pain/dysuria…we think UTI or pyelonephritis.
  …but what are we forgetting? We have the potential for COGNITIVE ERRORS.
REPEAT: A Lisa Sanders case walks in 27 yr. old Caucasian male

- Previous provider:
  - Labs (WNL) **New labs?**
  - Acupuncture
  - No other workup
    Ultrasound? GI referral?
- Here for a 2\textsuperscript{nd} opinion
  - Weight loss 100 pounds **RED FLAG**
  - Nausea/Vomiting
  - Fatigue

- **PMH:**
  - anxiety/depression, vitiligo-**FROM BIRTH?**
- **PSH:** none
- **Family hx:** none
- **DOUBTFUL**
- **Medications:** Zofran, marijuana for anxiety
Types of Cognitive Errors
(adapted from https://www.merckmanuals.com/professional/special-subjects/clinical-decision-making/cognitive-errors-in-

- Availability error: Recent experience
- Representation error: Probability with findings vs prevalence
Types of Cognitive Errors

• **Premature closure**: quick diagnosis, fail to consider other possible diagnoses, and stop collecting data

• **Anchoring errors**: cling to first initial thoughts
Types of Cognitive Errors

- **Confirmation bias** occurs when clinicians selectively accept clinical data that support a desired hypothesis and ignore data that do not (cherry-picking).

- **Attribution errors** involve negative stereotypes.
Types of Cognitive Errors

- **Affective error**
  avoiding testing that could “hurt” or be “embarrassing”
AVOID COGNITIVE ERRORS

• History/Physical
• Working diagnosis based on heuristics
• STOP. BREATHE. REFLECT. THEN...

• ASK
  – What else could it be?
  – What are the most dangerous things it could be?
  – Is there any evidence at odd with this working diagnosis?
Zebras in practice: Back Pain with a twist

• 32 yr old male
• Back pain
  – Moved to Oregon for insurance
  – Homeless in shelter
  – Can’t work due to pain
  – ER gives limited opioids
  – Diagnosed as sciatica
  – Can’t even go to PT
Zebras in practice: Back Pain with a twist

• #2 reasons for visit across America.
• Cognitive Errors (bias?)
  – Work up in ED?
  – Unreliable historian/language barrier?
  – Drug seeking?
  – Poor?

RED FLAGS:
- worsening exams
- persistent tachycardia and htn (from the back pain?)
Zebras in practice: Back Pain with a twist

• Remember the unexpected can happen.
• Work up of persistent HTN
  – Labs normal
  – 24 hour urine for catecholamines
Zebras in practice: Back Pain with a twist

- **Pheochromocytoma**
  - Fever than 20,000 cases yearly
  - Rare tumor that sits atop adrenal gland
  - Releases norepinephrine and epinephrine
  - Increases HR and BP

- **Prognosis**
  - Non-cancerous tumor with surgery, 95% survival
  - Cancerous, 50%
Zebras in practice: Unrelenting IDA

• 42 yr old female
• Hx of depression, anxiety and fatigue
• Fatigue workup
  – Ferritin 4ng/ml (<20 likely IDA)
  – Oral supplementation unhelpful
  – IV iron infusions
• Recheck of ferritin level post infusions
  – 3ng/ml
• Refer to HEME
Zebras in practice: Unrelenting IDA

- Patient’s daughter brought in for fatigue
- Ferritin also low
- Non-menstruating female
Zebras in practice: Unrelenting IDA

• Mom just got back from Panama with daughter. Mom was in the Peace Corps 20 years ago.
Zebras in practice: Unrelenting IDA

• Helminths
  – Parasitic Worms
  – Typically in stool that can be transmitted via food but primarily by walking barefoot where folks may have stooleed.
Your turn: pair and share activity

- What was your zebra?
  - Pearls of wisdom
  - If I could do it over again I would have…
Care for our zebra: The NP Role

- Thorough histories and physicals
- Avoid COGNITIVE ERRORS
- Judicious testing but questioning when patterns do not match traditional presentation
Care for our zebra: The NP Role

• Connecting patients to specialists

• NORD (National Organization of Rare Disorders)
  – Database of 1200 diseases
  – Orphan Drug Act/Access medications
Care for our zebra: The NP Role

• Zebras are horses too
  – Advocating for general care
    • immies
    • screening
    • health promotion

  – Safety planning
  – Knowledge: https://rarediseases.org/for-patients-and-families/information-resources/physician-guides/
Thank you for your attention and participation!
References


FACT SHEET