

POST-PBL WRAP-UP

PROBLEM-BASED LEARNING:

**A 26-year-old female with episodic flushing,
abdominal pain, diarrhea, and dizziness**

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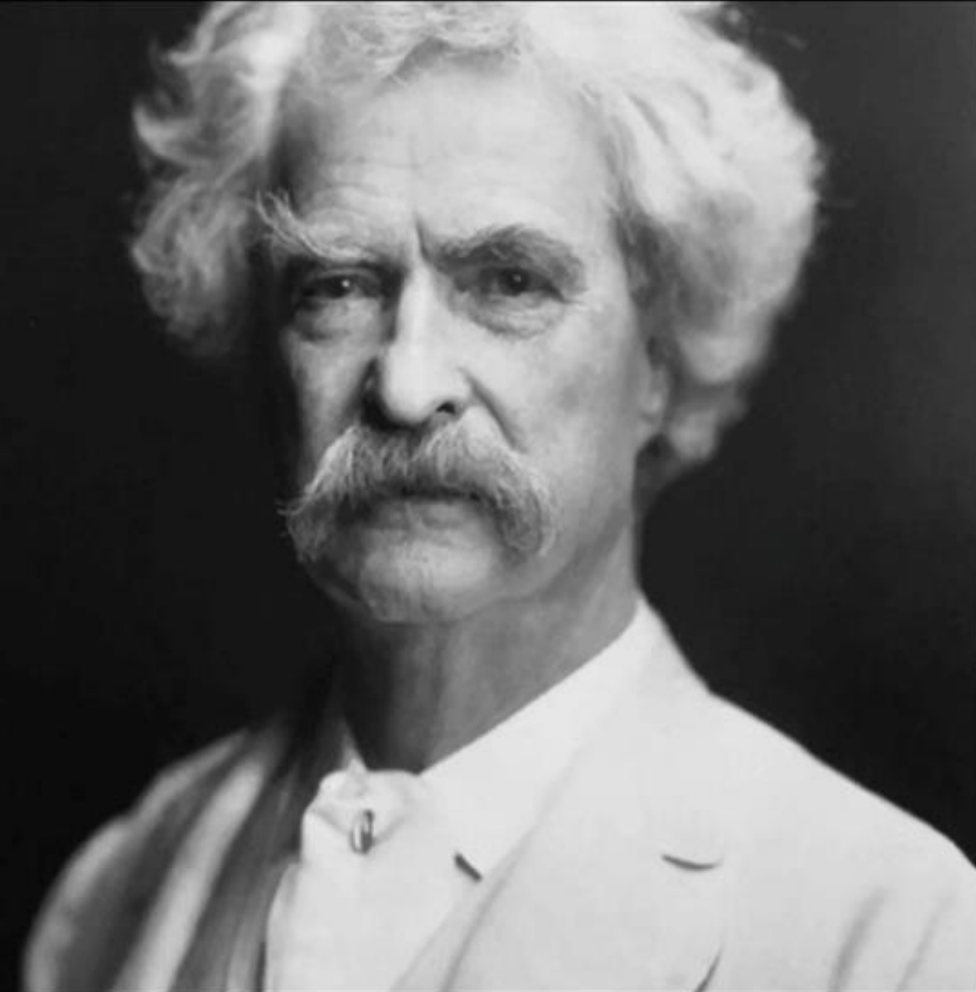
Brigham and Women's Hospital

Harvard Medical School

POST-PBL WRAP-UP

It ain't what you don't know
that gets you into trouble. It's
what you know for sure that
just ain't so.

Mark Twain



HISTORY

- Alexa is a 26-year-old female with many complaints including intermittent cramping abdominal pain, diarrhea, flushing, and dizziness for 6 years. She states, “I have seen all the specialists (internal medicine, cardiology, gastroenterology, endocrinology, allergy, and my chiropractor) and nobody knows what to do with me.” My doctors tell me that these symptoms are due to IBS and panic attacks. She is seeing you for another opinion
- Symptoms have progressively become more frequent and more severe over the past couple of years. On a few occasions, when her flushing has been at its worst, she has felt so lightheaded that she has almost passed out. Although the history does not suggest a consistently identifiable provocation, some flushing episodes seem to have been triggered by exercise, alcohol, spicy foods, and emotional events
- She is carrying a stack of records for you to review

PAST EVALUATION

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- **Internal Medicine:** chemistry panel, thyroid testing, CBC w/o diff, ESR, CRP, ANA, and CXR - all normal except for a mildly elevated alkaline phosphatase
- **GI** - normal endoscopy and colonoscopy. No biopsy was performed. A proton pump inhibitor was prescribed for two months without benefit
- **Cardiology:** EKG, halter monitoring, and cardiac echo – normal; Tilt test – negative
- **Endocrinology:** Normal 24-hour urinary 5-HIAA. Normal serum vasoactive intestinal polypeptide.
- **Allergy (ENT):** Negative serum IgE food panel and multiple positives on dilution-titration environmental ST. Treatment with H1 and H2 blockers (hydroxyzine and famotidine) was too sedating for the patient to tolerate.
- **Chiropractor:** prescribed a trial of the FODMAP diet. After it failed to provide benefit, he told her that she probably suffered from “histamine intolerance,” and then put her on a low-histamine diet, which was also of no benefit.

ADDITIONAL HISTORY (part 2 of 2)

- **REVIEW OF SYSTEMS:**
- **SKIN** – Episodic flushing; generally lasts a few hours and is not accompanied by sweating. Life-long freckled skin.
- **GI** — positive for intermittent nausea, vomiting, diarrhea, and bloating associated with abdominal cramping pain
- **Resp** — negative
- **CV** — Few episodes of lightheadedness preceded by palpitations and at times a “near syncope feeling” that have no consistent trigger, sometimes after certain foods, sometimes when exposed to heat or under stress.
- **Rheumatologic** — some musculoskeletal aches; denies joint hypermobility
- **Neuropsychiatric** — headaches, fatigue, and difficulty concentrating
- **PAST MEDICAL HISTORY:** No history of COVID (fully immunized)
- **DRUG ALLERGY:** Narcotics (hives)
- **ENVIRONMENT:** No pets or smokers
- **SOCIAL HISTORY:** Alexa works from home
- **FAMILY HISTORY:** Noncontributory

PHYSICAL EXAM

- GENERAL: Well developed in no acute distress.
- VITAL SIGNS: sitting: BP 115/70; P-76; standing: BP 108/62; P-82
- HEART – Regular Rate & Rhythm
- LUNGS - Clear
- SKIN – scattered brownish red freckles on the back and upper extremities; questionable urtication with stroking
- MUSCULOSKELETAL EXAM – normal joint mobility (normal Beighton score)

IMPRESSION

1.

2.

3.

4.

EVALUATION

1.

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INITIAL LAB RESULTS (within 48 hours)

- **BASELINE SERUM TRYPTASE LEVEL = 18** (*normal <11*)
- **CBC: H/H = 11/33, mcv = 92; platelets = 145K; WBC = 4,400 (nl diff)**
- **CMP: mildly elevated Alk Phos (160 U/L) with normal transaminases**
- **Skin test: Skin test negative to multiple foods and spices**
- **Negative serum IgE to alpha-gal**

IMPRESSION

- 1. UNDIFFERENTIATED MAST CELL DISORDER**

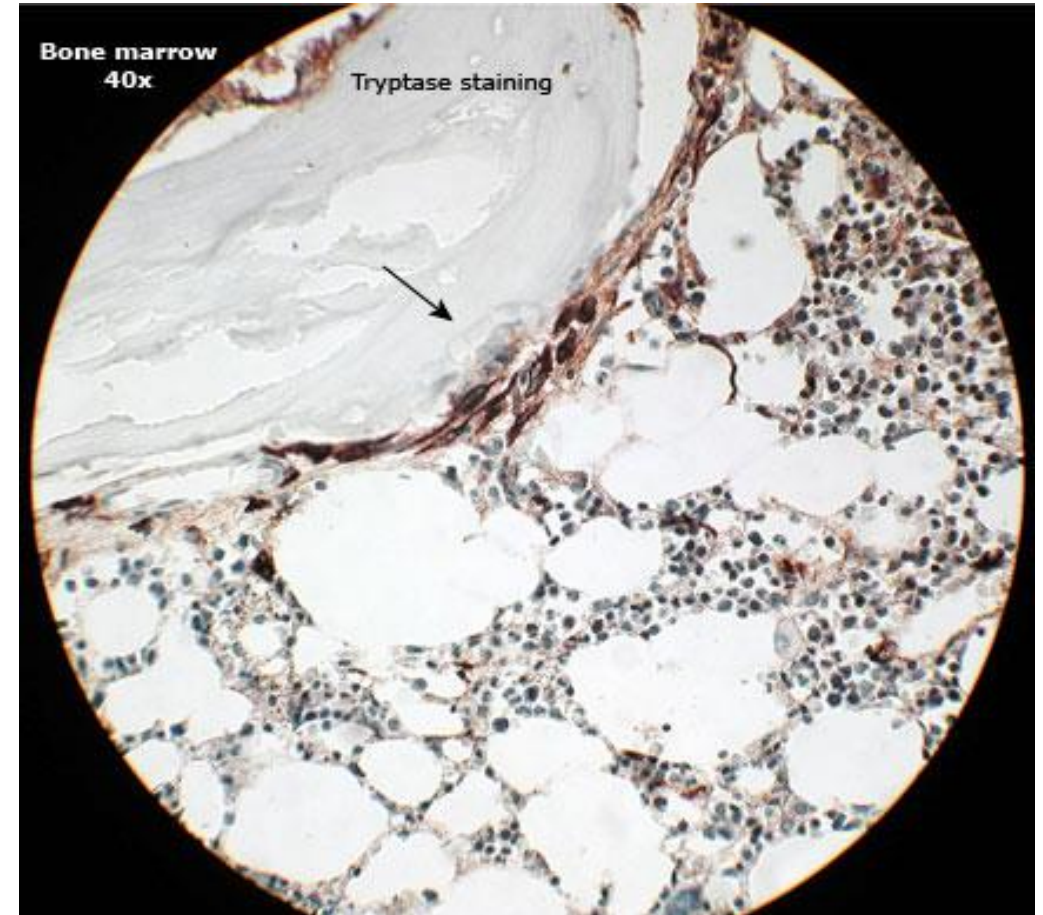
FURTHER EVALUATION

- 1.**

- 2.**

FURTHER LAB RESULTS

- **SERUM TRYPTASE LEVEL = 22**
- **SKIN BIOPSY:** positive immunohistochemical staining for tryptase and KIT
- **PERIPHERAL BLOOD:** *KIT* D816V DIGITAL PCR TEST - POSITIVE
- **BONE MARROW BIOPSY:**
Tryptase-stained bone marrow sections revealed multifocal, dense aggregates of > 15 mast cells. Bone marrow sections stained positive for tryptase, CD2, CD25, and CD30



FINAL DIAGNOSIS:

INDOLENT SYSTEMIC MASTOCYTOSIS

FINAL RECOMMENDATIONS:

- 1.**
- 2.**
- 3.**