03/14/24 Morning Report with @CPSolvers

"One life, so many dreams" Case Presenter: Khashayar (@) Case Discussants: Rabih Geha (@rabihmgeha) and Shreyas Nandyal (@shreyas_rn)

CC: 24 M referred for diarrhea significant weight loss

HPI: Symptoms started since he was 8 years old → Watery diarrhea, fatigue, anemia, episodic jaundice → Changed severity worsened since 2 mo ago; weight loss 20 Kg last year, hypomagnesemia, hypocalcemia and hypokalemia.

PMH: Celiac (10 years old not documented) Mucocutaneous candidiasis, GI candidiasis, Delayed puberty at age 18, T1DM, Adrenal insufficiency, Hypoparathyroidism, Hypothyroidism

Meds: Calcium carbonate, Calcitriol, FLudrocortisone, Prednisone, levothyroxine.

Fam Hx:

Soc Hx:
No alcohol
No smoking

Health-Related Behaviors:

Allergies: no allergies

Vitals: T: afebrile **HR:** 80 **BP:** 90/60 **RR:** 22 **BMI** 15.5
Exam:
Gen: Polyuria, polydipsia, difference in testes size dysphagia w/ solid foods.
HEENT: hyperpigmentation buccal mucosa
Abd: no pain

Notable Labs & Imaging:
Hematology:
WBC: 1.6 Hgb: 8.6 Plt: nl
Chemistry:
Na:131 K: 2.7 Cl: HCO3: 16 BUN: Cr: 1.9; Glucose: nl (multiple episodes of hypoglycemia); Ca:4 Mag: 1 Albumin: 2.5 PH: 7.24 Pco2: 42 PTH 2.6 Anti TTG IgG & IGA (neg) ACTH: 699 TSH: 2.7 T4: 3.5 calprotectin 0.3 µg/g 24 hr urine Na 54 meq/day

Imaging:
US abdomen and pelvis: 21 mm gallbladder stone, liver, spleen, hepatic portal vein nl, pancreas nl, kidney reduced size (R 84 L 85 mm) with multiple echogenic foci (probably kidney stones), no free fluid.
MR enterography nl.
Pathology report: gastric (mild chronic gastritis no metaplasia dysplasia atrophy, no H.pylori) and intestinal mucosa (unremarkable villous structure preserved no intra epithelial hyperplasia)

Dx: Suspected **APS-1** (autoimmune polyglandular syndrome type 1)

Problem Representation: 24M with chronic diarrhea and multiple endocrinopathy since childhood presents with weight loss, anemia and multiple electrolyte abnormalities

Teaching Points (Francisco):
Diarrhea + weight loss + anemia + electrolyte abnormalities + jaundice (episodic) + long standing
Chronic Diarrhea: inflammatory (IBD, chronic infection), secretory (loss of water, decreased absorption), osmotic
Anemia + diarrhea = malabsorption (GI or biliary tract)
Diarrhea -> hypoMG -> hypoCa
Chronic jaundice is rare, profound filter given the rarity
Jaundice + anemia -> liver disease (macrocytic), hemolysis
Endocrinopathy (thyroid, parathyroid, adrenal) + multiple = autoimmunity, polyglandular and infiltrative dx (Tb and histo)
Episodic jaundice: false localizing of jaundice - distractor (sepsis and heart disease - congestive hepatopathy) - can be a clue to a genetic disease like Gilbert disease (common in Middle East) or recurrent intrahepatic cholestasis
"Turning yellow in arabic means not feeling well"
Dysphagia: obstruction (starts on solids and is progressive) vs. motility (solids + liquids)
Dysphagia can cause nutritional deficiency, what about the opposite - think about Plummer Vinson (bidirectional)
Cytopenias is a cause of consequence of nutritional deficiency?
Increased ACTH in response to the adrenal insufficiency (pituitary is not affected)
Primary adrenal insufficiency (hyperpigmentation, hypoTa w/o tachycardia, high urine Na, high ACTH)
Celiac antibodies NEG: adherent to the diet, IgA deficiency, does not have the disease
hypoPTH causes hypoCa and nephrolithiasis (Ca wasted in the kidney)
Chronic diarrhea in a systemic issue, GI involvement is a consequence and not cause
Polyendocrinopathy: genetic (autoimmune) or acquired (mass or infiltration)
Autoimmune polyglandular syndrome: presence of adrenal insufficiency (always) and are classified by a immune deficiency
Type 1 (hypoPTH and adrenal insufficiency and chronic mucocutaneous candidiasis, measure IFN antibodies and gene sequence of AIRE) and 3 (IPEX - infancy, immunodeficiency and atopic in the spectrum of Job) have immunodeficiency
Type 2 (schmidt syndrome): most common, middle age, lacks immunodeficiency