10/09/23 Rafael Medina Subspecialty VMR with @CPSolvers

"One life, so many dreams" Case Presenter: Christopher Coe (@CCOE2990) ***Case Discussants****: Dr. Patel (@ArpanPatelMD)*

CC: Worsening fatigue

HPI:
33F seen in hepatology clinic. She is a refugee w/o records coming from Europe who presents w/ worsening fatigue and depression for the past 1 year. Saw a doctor in Poland who performed abd. US which revealed "large spleen".
GI hx: no N/V, melena/hematochezia, confusion. Non jaundice or confusion. No pruritus.
ROS: worsening depression. No suicidal ideation.

PMH:
Obesity
No PSH
Issues conceiving for last 18 months

Meds:
Sertraline 50mg qd

Fam Hx:
Sister: leukemia.
Mother /father: obesity, hyperlipidemia
Mother: Breast cancer

Soc Hx:
Secretary for mechanic office in Eastern Europe.
3-4 glasses of wine per week.

Vitals: T: HR: 73 **BP:** 101/64 **RR: SpO2** 98% on room air. Weight: 198Lb (BMI 30.1)
Exam: Gen: no acute distress. Normal affect
HEENT: no conjunctival icterus.
CV: nl, **Pulm:** nl; **Abd:** obese abdomen with central adiposity, no fluid wave, LUQ fullness. No hepatic bruit.
Neuro: AAO x4. No asterixis. Fine postural tremor.
Extremities/skin: no jaundice, no spider angiomata, no palmar erythema, washed out nails (thin brown end on nails), no sarcopenia or temporal wasting. No peripheral edema.

Notable Labs & Imaging:
Hematology:
WBC: 4800 Hgb: 11.7 (MCV: 95) Plt: 68k
Chemistry:
Na: 134 K: 3.9 BUN: 15 Cr: 0.57 glucose: 95. AST: 75 ALT: 65 Alk-P: 71 T. Bili: 0.6 Albumin: 3.3. INR 1.3
Advanced labs:
Hepatitis serologies negative. CMV/ EBV (-). Ferritin 68, Tsat 28%. A1AT genotype: MM. Ceruloplasmin 4 (nl: > 14), Serum copper 61.8 (80-155). ANA/ anti-SM/ AMA negative. Total IgG 1005. 24h urine copper: 81 mcg/day (nl < 35). Non ceruloplasmin bound copper: 49.2 (nl < 10-15 mcg/dL)
Imaging:
Abdominal US: heterogeneous echotexture compatible with steatosis. Nodular liver contour. Moderately enlarged spleen (17.2cm). No ascites
Liver biopsy:
mild cholestasis and steatosis. Trichrome stain: mild portal fibrosis. Rhodamine stain: red brown granules. 894 mcg copper/gram of dry weight (nl < 50)
No Kayser- Fleischer rings on Ophthalmology evaluation

Final dx: Wilson's disease

Problem Representation: 33yF w/ obesity who recently moved from Europe p/w worsening fatigue, depression and recent US finding of splenomegaly. Labs notable for thrombocytopenia, low ceruloplasmin and low serum-copper. Liver biopsy consistent w/ copper infiltration.

Teaching Points (@Noah_Nakajima):
Splenomegaly: Increased platelet consumption or increase in pressure on the spleen.
➞ Portal hypertension jumps out: can be from pre, intrinsic, or post hepatic.
Fatigue in the context of liver disease: Cirrhosis, acute viral hepatitis, primary biliary cholangitis.
Time-frame is key. Thought-exercise, acute splenomegaly + fatigue ➞ EBV is a strong possibility.
Physical examination:
Cirrhosis: spider angiomata, palmar erythema, signs of encephalopathy.
➞ Asterixis can be due to toxic metabolic encephalopathy. Ask patient to stop traffic or do the milkmaid sign.
Portal hypertension: ascites, caput medusae, splenomegaly.
Terry's nails: washed-out nails with thin brown-red line on the ends ➞ liver dz or metabolic syndromes.
DDx: Steatotic liver disease ➞ Metabolic Dysfunction Associated Steatotic Liver Disease +/- MetALS (alcohol).
Labs & Imaging:
Nodular liver *not always* cirrhosis! Congestive hepatopathy, nodular regenerative hyperplasia, etc.
However, with other clues (thrombocytopenia, low albumin), c/f cirrhosis. Important to work up anemia.
Steatosis on the US: Fat in the liver, from metabolic syndrome, alcohol intake, medication-related, idiopathic.
Don't fall for the search satisficing trap! If the patient has risk factors for MASLD or MetALS, it does not mean that they will only have this diagnosis. You need to send hepatitis serologies, hemochromatosis workup, autoimmune if warranted.
MASLD Dx: Metabolic syndrome + Steatotic liver disease (imaging or fibroscan).
Cirrhosis Dx: Imaging + thrombocytopenia + likely etiology + excluding ddx.
Wilson's disease: Ceruloplasmin can also be low in liver disease. Clinical picture is paramount for the dx. Possible advanced labs is urine copper and ophto evaluation.
A1AT genotype: The most concerning for liver disease is ZZ.
Fibroscan: Useful for quantifying the liver damage, evaluating progression and risk stratifying for endoscopy (Baveno VI criteria).
Young age: Makes us more concerned for other etiologies than MASLD in this patient ➞ Value in working up Wilson's disease.
There is no cookie-cutter recipe! Individualize diagnostic and management decisions to the patient.
Wilson's dz can present as lean steatotic liver disease.
Highlights: Clinical dx of cirrhosis does not depend on classic physical signs. Framing of young patient pushes us to investigate further. Patients with liver dz do not always have 1 diagnosis - don't be search satisficing.
Close follow-up is key to diagnose and manage effectively those cases!