9/21/23 Morning Report with @CPSolvers

"One life, so many dreams" Case Presenter: Mark Heslin (@Mark_Heslin) Case Discussants: David Serantes (@davserantes) and Rabih Geha (@rabihmgeha)

CC: 62 yo male presenting with 2 days of **generalized weakness**, Lower Extremity **numbness**, 1 month of **polydipsia and polyuria**

HPI: No healthcare encounter since age 20. Difficulty in obtaining history from patient. Information obtained from a relative due to patient inability to provide coherent history owing to decreased orientation.

PMH: unknown

Meds: unknown

Fam Hx: unknown family history

Soc Hx: retired, lives with brother

Health-Related Behaviors: not known

Allergies: None

Vitals: T: afebrile **HR:** nl **BP:** nl **RR:** 5L nasal cannula
Exam:
CV: normal
Pulm: crackle right lower lobes
Abd: normal;
Neuro: diffuse myoclonus, carpopedal spasm while measuring BP, Not oriented to time and place
Extremities/skin: ⅘ strength all extremity, mild hyporeflexia patellar tendon bilaterally, no edema

Notable Labs & Imaging:
Hematology:
WBC: 15.7k (predom neutrophilia) Hgb: 15.7 Plt: 357
Chemistry:
Na: K: 5.4 Cl: 96 BUN:45, Cr: 1.86, glucose: 486, Ca: 7.5, Mag: 2, Phos 5.2
AST: 90, ALT:42, Alk-P: 67, T. Bilirubin 0.7, Total Protein: 7.5, Albumin: 3.5, CK 1941, UA normal BNP 35 Urinary toxicology & Serum Ethanol negative Troponin HA1c 9%, ABG - Ionized calcium of ?, Anion Gap - 9, Vit D nl, low PTH (5)
TSH normal, HIV & RPR non reactive, Hepatitis serology- normal, Transferrin percent saturation high, Ferritin high
Imaging:
CXR: right lower lobe opacity
POCUS: globally reduced EF (36%)
Echocardiogram:
Nuclear stress test: LEF 36%, no evidence of reversible ischemia
RUQ US hepatomegaly, L liver lesion
CT head negative
MRI brain: suspectic ischemia in frontal areas?
Calcium gluconate was given, ceft was given for CAP
MRI liver: Increase uptake liver & spleen without cirrhosis, Hepatic haemangioma on left side without HCC features
Genetic study: Homozygous for Hereditary Hemochromatosis mutation C282Y
Dx: Hereditary Hemochromatosis

Problem Representation: A 62yM p/w weakness, numbness, polydipsia & polyuria. PE notable for diffuse myoclonus, Trousseau sign & diffuse crackles in RLL w/ hypoxemia. Labs notable for leukocytosis, elevated CK & signs for multiple endocrinopathies (hypocalcemia, low PTH, hyperglycemia (A1C 9%) & elevated iron studies). Imaging shows RLL opacity, HFrEF & hepatomegaly.

Teaching Points (Francisco):
2 days of generalized weakness and lower extremity numbness: assess the time and how they are related -> neurological event with **rapid evolution** (think about electrolytes)
3's in Common Neuropathy: 1) Symmetric 2) sensory predominant and 3) slowly progressive -> if the 3 are not present, neuropathy is atypical or there is a mimicker
Atypical neuropathy -> 2 data points -> reflexes (if non-existant think GBS until proven otherwise) + glucose (DM)
3 dimensions:
- **Metabolic**: myoclonus, carpopedal spasm -> electrolyte problem -> hypocalcemia (get ABG + EKG)
- **Hypoxemic respiratory failure**: assess signs of infection and volume overload -> CAP (consolidation + leukocytosis) -> think about atypical organisms (extrapulmonary spread)
- **Muscle:** CK elevation and AST/ALT >2:1 -> give fluids

Causes of hypoCa: vitD def, PTH def or chelation (latter common in acute)
HypoPTH: hypoMg, surgery (neck), genetic, autoimmune
Multiple endocrinopathies (DM and hyperPTH):
- Pituitary adenoma
- Hemochromatosis (hepatomegaly + iron overload + dilated cardiomyopathy)
- POEMS
- Wilson's Disease

Iron overload causes: transfusions, liver disease (alcohol), hematologic disease (MDS), genetic (low penetrance - 20%, dx of exclusion even if genetic test +)
Phlebotomy testing to distinguish from hereditary (don't develop anemia) to non hereditary.