8/10/23 Morning Report with @CPSolvers

***"One life, so many dreams" Case Presenter**: Tansu Eris (@drtansue) **Case Discussants**: Ayesha Ghoto (@AyeshaGhoto) and Rabih Geha (@rabihmgeha)*

CC: 40yF w/ fatigue and blurry vision

HPI:
40yF w/ fatigue & blurry vision for years, symptoms life-limiting while performing daily chores
11y ago: Derm clinic for spontaneous facial freckling → ferritin 964
9 y ago: fatigue w/ gradual onset + constant blurry vision on both eyes; early phase of b/l cataract
2 y ago: fatigue improved; Ferritin 1218; 6d after new treatment stomach aches; husbands reports she vomited + LOC after, contracting upper and lower extremities → ED: still LOC + tonic clonic movements; sedation control → intubation; EEG, CT brain, whole body imaging normal → Status epilepticus
Complaints subsided after stopping the medication

PMH:
none

Meds:
none

Fam Hx:
Father died due to stroke at 62
Aunts+cousin from maternal side: b/l cataract
Sister+niece b/l cataract (age 16, 30))

Health-Related Behaviors:
none

Allergies:none

Vitals: T:36.7 **HR:**85 **BP:**100/60 **RR:**20
Exam:
Gen: no acute distress, neck supple, no LAD, conjunctiva pale
CV: regular rhythm
Pulm: clear to auscultation
Abd: non tender, no mass or organomegaly
Neuro: nl, alert oriented w/ normal speech
Extremities/skin: no edema, facial freckling, pallor

Notable Labs & Imaging:
Hematology:
WBC: nl → 7.5 Hgb: nl → 8.3 (MCV 61.5) Plt: nl → Ferritin 1399 → peak >2000 (2 y prior), TIBC: 492
TSH, Folate, Vitamin B12 nl
Chemistry: CMP: normal, Coags normal
11y ago: Hep panel Hep A, B, C negative, HIV neg, Hemoglobinopathies neg (electrophoresis); genetic testing for Hereditary hemochromatosis (HFE gene) positive for H63D mutation (homozygote) → Phlebotomy induced iatrogenic iron def. anemia → after fatigue
2y ago: Deferoxamine was started → Hereditary hemochromatosis: Neuro hereditary hemochromatosis less likely because no signs of iron accumulation in whole body imaging → Deferoxamine discontinued
Lack of evidence for iron overload + IDA + b/l cataract + hyperferritinemia
Positive FTL gene mutation (Chr. 19q)

Final dx: Hereditary hyperferritinemia cataract syndrome (HHCS)

Problem Representation: A 40 yF w/ a h/o hereditary hemochromatosis (H63D) homozygous mutation and deferoxamine regimen presented to the hospital w/ fatigue and long-standing b/l cataracts. Labs were remarkable for microcytic anemia and hyperferritinemia. Multiple maternal family members also had b/l cataracts.

Teaching Points (Marino):
-Fatigue + blurry vision: fatigue is a very non specific symptom but blurry vision can help us to narrow down on diagnostic possibilities.
-Everytime we get a patient with fatigue, we must ask questions such as: Is there fatigue at rest or with activity?
-Fatigue: hormones, iron deficiency. Could also be a loose description of exercise intolerance.
-Fatigue with exertion ddx: neuro/MSK/cardiopulmonary system/anemia.
-Blurry vision: fussy quality of image. Don't confuse it with loss of vision although it is interchangeable between patients.
-Skin findings + neuro findings + blurry vision: Neurocutaneous disorders should be included in the differential.
-Bilateral cataracts: steroid use, osmotic damage, UV radiation.
-**Medication-triggered seizure**: Many medications can predispose to seizures in patients with lower seizure threshold.
-Very rare to see hemochromatosis in a 40 year old female: periodic bleeding from periods.
-Causes of hyperferritinemia: Inflammatory or non-inflammatory.
-Disease presentation in multiple family members should guide us towards the genetic bucket.
-Microcytic anemias: IDA, anemia of chronic inflammatory disease, thalassemias and lead poisoning.
-Iron overload triggers in patients with hemochromatosis: HIV, HBV, HCV and alcohol.
-Hemochromatosis diagnosis is made after seeing improvement with treatment. Genetic testing only gives us clues about which mutation is present. Only 20% of patients have phenotypic expression. Listen to your patient and look for signs of improvement.
- Hereditary hyperferritinemia cataract syndrome (HHCS): FTL gene on chromosome 19q; 30 variants of FTL gene; autosomal-dominant pattern; benign hyperferritinemia)