10/13/26 Neuro VMR with @CPSolvers

"One life, so many dreams" ***Case Presenter****: Kaleem(@)* ***Case Discussants****: Helen Shi(@) & Aye(@)*

https://clinicalproblemsolving.com/present-a-case/

Scribing (Sarah B):

CC: 15 yo male with referred to neurology for sudden **painless loss of vision** in the **right eye** for 2 days.

HPI: Vision loss was **maximal at onset**, occurred within 2 minutes. No floaters, no redness, no trauma, no flashes, no diplopia.

ROS:
Positive for **low grade fever** and **arthralgias**.

PMH:
5 yo: Febrile seizures
7 yo: diplopia and head posturing. Possible stroke, lost to follow-up.
10 yo: Painful joints and rash–inconclusive biopsy
11 yo: Recurrent fever, abd pain, myalgias, weight loss. CTA w/medium artery aneurysm. Polyarteritis nodosa diagnosis.
12 yo: capsuloganglionic infarct

Meds:
Cyclophosphamide
Mycophenolate
Rituximab
IVIG
Methotrexate

Fam Hx:
Consanguinity in parents
Juvenile Idiopathic Arthritis in sister

Developmental Hx:
Cesarean delivery
Met developmental milestones
Breastfed until 6 months age
Immunizations UTD
BCG scar present

Allergies:
none

Vitals: T: 98F **HR**: 80 **BP**: 125/18 **RR**: 16 **Sat**: 98% **BG**: 102
Exam: Gen: young male, comfortable in chair with pallor/rash present
HEENT: **CV**: nl **Pulm**: nl **Abd**: nl
Neuro: oriented/alert. R eye finger counting only. Fundoscopy with pale retina and macular cherry red spot. L eye normal. R RAPD. **Residual** R sided weakness. Power ⅘ in RUE/RLE though able to walk unaided.
Extremities/skin: Net-like rash around body with irregular branch-like skin discoloration. No clubbing/cyanosis or jaundice.

Notable Labs & Imaging:

Hematology:
WBC: 7.3K Hgb: 9.7 Plt: 157 MCV: 65

Chemistry: Normal

Advanced Labs
Thrombophilia screen: nl
AST: nl ALT: nl ALP: nl T. Bili: nl T. protein: nl Alb: nl
ESR: 47 CRP: 50 TIBC low Ferritin: normal
HBsAg, HCsAb, RPR, HIV, Blood cx: negative
ANA/RF/anti-dsDNA/Lupus AC/p- c-ANCA/ENA/pathergy: negative
UA: negative

Imaging:
Echo: 60% EF
CT Brain: normal
MRI Brain: chronic lacunar L thalamic infarcts, frontal lobar infarct

Dx: Adenosine deaminase 2 deficiency, treated with etanercept and methotrexate with folic acid supplementation

Problem Representation: 15 year old male with significant history of arthralgia, medium vessel arteritis resistant to immune suppression presents with painless R eye vision loss found to have rash and elevated inflammatory markers.

Teaching Points (Kwame):
Sudden onset vision loss -CRAO,Retinal detachment, Amaurosis fugax

Monocular vision loss did it happen with 4 and half hours?
Ischemic optic neuropathy -arteritic example GCA or non arteretic

Bartonella fever night sweats abdominal pain, vision loss but would have pain associated with it. Infective endocarditis could lead to a septic emboli in the heartland would express low grade fever..

Due to chronicity an infection is less likely, thinking more of an autoimmune condition.

RAPD suggest there's optic nerve involvement but a glaucoma cataract etc can prevent light from reaching the midbrain.Initial CRAO may have normal fundoscopy initially.

After negative autoimmune panel
Pathogenetic mutation in ADA 2 gene -treatment etenerceptsupplementd with folic acid