9/16/26 Morning Report with @CPSolvers

"One life, so many dreams" ***Case Presenter****: Kaleem(@kaleem_it.is - IG)* ***Case Discussants****: Sharmin (@Sharmin) & Reza(@)*

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

Scribing (Praveen):

CC: 26 M % worsening hand tremors and leg weakness and frequent falls

HPI:

Apparently well 3 years ago. No longer able to eat with hands gradually and developed leg weakness.

Urine copper high -> Dx as Wilson's.

Progressive tremors, leg weakness with inability to walk [L > R], slow movements, dysarthria and dysphonia.

ROS: no neck stiffness, headaches, sensory Sx, no sore throat, no SOB / cough, palpitation, orthopnea, urinary / bowel Sx, no rashes, and otherwise negative.

PMH:
Wilson's Disease

Meds:
Penicillamine
Zinc Sulphate

Fam Hx:
Sister with similar symptoms not on Rx

Social Hx:
Nil Relevant

Vitals: T: afebrile **HR**: 80 **BP**: 123/85 **RR**: **Sat**: 99%

Exam: Gen: Lying comfortable, AO x 3

HEENT: nl **CV**, **Pulm**, **Abd**: nl

Neuro: Optho: eyes with no Kayser-Fleischer rings. No nystagmus.

Motor: Rigidity, power ⅘ RUE, 4/5 all other limbs. Reduced reflexes, no clonus. Plantar response upgoing bl.

Coordination: Tremor provoked by movements. Dysdiadochokinesia, dysmetria. Broad based and slow gait, favoring L side. Hypophonic and dysarthric speech, reduced facial expression.

Notable Labs & Imaging:

Hematology: CBC - nl

Chemistry: Serum Electrolytes, LFT and Coagulation profile - wnl

UA : nl Cr : 1.29

Serum Cu, Ceruloplasmin, 24hr Urinary Copper - nl

Imaging

CT Brain - nl

CT Abd Pelvis - nl

MRI Brain - nl

EMG - nl NCS - nl

Liver USG : Coarse and increased hepatic echotexture suggesting diffuse hepatocellular parenchymal disease

Clinical Course: Symptoms progressed. Genetic testing revealed a heterozygous variant of GIGYF2 gene associated with Parkinsonism Type 11 with AD or sporadic inheritance. Patient on treatment and slowly improving with monitoring.

Sister was also evaluated later, found positive for the same mutation and treatment started with clinical improvement.

Diagnosis : GIGYF2 - Associated Parkinson Disease Type 11 / Wilson's Disease

Problem Representation: 26 year old male with known Wilson's disease presents with a gradual worsening UL tremors and LL weakness over 3 years. Similar symptoms present in sister. Examination revealed parkinsonian features.

Teaching Points (Mattia)

When multiple findings: choose one to focus on, the one you are most comfortable with, in the first place

Neurological symptoms:

Leg weakness: objective findings?(eg standing up from chair?) Is it possible to localize a lesion explaining it?

- Muscle
- Neuromuscular junction
- Nerve

Tremor: Spasm? Myoclony? Parkinsonism?

Other symptoms associated?

- Rigidity?
- Sensory neuropathy?
- Cerebellar symptoms?

Reevaluate past medical history:

Is the Wilson disease (or any major diagnosis) explaining all the findings?

Otherwise risk of anchoring bias.

Question the initial diagnosis if discordance seems to appear through the diagnostic journey

Weakness + dysphagia+ dysphonia:: evaluate possibility of bulbar symptoms.

MRI in evaluation of the symptoms:: objective findings (eg demyelination) or imaging negative?

Parkinsonism in young patients: need for evaluation for genetic causes

When multiple neurologicals systems involvement genetic and metabolic problem is possible, evolving in a chronic neurodegenerative disease