1/30/26 Morning Report with @CPSolvers

"One life, so many dreams" ***Case Presenter****: Hee Mun* ***Case Discussants****: Dr. Ravi (@rav7ks) & Prof. Reza*

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

Scribing (Krithika)

CC: Hyperkinetic movement disorder

HPI: A 22 y/o female with a 3 year h/o action induced **jerks** (eg brushing teeth) **and tremor** while carrying drinks, **urinary copper elevated twice**. Gabapentin was ineffective, clonazepam initially helped but months later she developed **fatigue, HTN, anemia, peripheral edema.**

ROS (-): headaches, hallucinations, preceding infection or toxin exposure, weakness, sensory loss, diplopia, dysarthria, urinary incontinence, tongue biting, fever, weight loss, night sweats, rash or oral ulcers

PMH: none

Meds: OCPs

Fam Hx:no family history of tremor, epilepsy in half brother

Social Hx: no smoking/ alcohol

Health-Related Behaviors: no alcohol, smoking history

Allergies: nil

Vitals: T: 36.8 **HR**: 78 bpm **BP**: 150/86 **RR**: 14cpm **Sat**: 99% @RA
Exam: Gen: alert **CV**, **Pulm**, **Abd**: nl
HEENT: No JVD, no Kayser-Fleischer rings, no jaundice
Neuro: CN 2 to 12- intact, No dystonia, rigidity,or bradykinesia,
Tremor: jerky and irregular with minimal dysmetria manifested as slight difficulty with ftf testing and tandem gait. Postural/action tremor of outstretched hands (jerky and irregular) with stimulus sensitive myoclonus
Brisk and symmetric reflexes, neg Babinski, Archimedes spiral-difficulty drawing with heavy pen pressure and irregular sudden jerky breaks.

Notable Labs & Imaging:

Hematology:
WBC: wnl except mild eosinophilia [AEC ?]
TSH: nl LFTs: nl
No acanthocytes on thick blood film- neuroacanthocytosis ruled out

Chemistry:
Autoimmune/vasculitis panel neg, Cr: 2.3 BUN: 22.2 Albumin < 3
ESR:81 , Egfr-26, Copper, Mg: nl Urinary copper: high(135,196) Ceruloplasmin: nl
UACR-3398MG/G(nephrotic range)
Serology-HIV, hepatitis panel: neg
EEG/MRI Brain/DaTscan-nl
Renal biopsy- Classic FSGS with severe podocyte injury, segmental and global sclerosis, secondary tubular damage from proteinuria and no immune complex disease. Some collapsing features but not meeting criteria for collapsing FSGS.

Genetic testing prompted by young age, myoclonus with renal failure, and FSGS on biopsy→SCARB2 mutation(autosomal recessive), progressive myoclonic epilepsy type 4

Rapid progression to ESRD→CAPD→living donor kidney transplant with normalized creatinine and resolved nephrotic syndrome.
On Tacrolimus/MMF/prednisolone →progressive myoclonus and worsening balance(likely tacrolimus toxicity)

DX- ACTION MYOCLONUS-RENAL FAILURE SYNDROME(AMRFS)

Problem Representation:22 year old female with hyperkinetic movement disorder with associated fatigue, hypertension, anemia and peripheral edema.

Teaching Points (Prof Eugene <3)

Hyperkinetic movement disorder:
-Explore background, what makes it worse?, meds?

Clues from Non-neurologic medical issues:
-**anemia, peripheral edema, HTN** -> may point away from only primary neurologic disorder, suggesting multisystem disease.(absence or deficiency of a substance or excess)
-**Anemia**: explore iron studies for IDA,Cu def., zinc excess etc.
-Edema: coupled with the movement disorder, consider TSH. Remember Liver, Heart, Kidney
-HTN: odd at her age hence explore sec causes (e.g., osa, renal artery stenosis, Fibromuscular dysplasia, pheochromocytoma
-**copper in urin**e: normal pathway should be excretion through bile. Presence in urine may suggest defect in loading copper to ceruloplasmin (aka. wilsons?) hence not incorporated in bile.

What kind of movement disorder:
-From video we see an overlap between action and intention tremor/myoclonus.
-Type of tremor/ underlying myoclonus does not suggest primary cause, rather either metabolic/ toxins or structural.
-A metabolic cause that may fit clinical picture may involve the kidney
-Still important to explore toxin affecting both kidney/ brain (image negative)

Labs
-nephrotic range proteinuria, low albumin
-high urine copper (cu can be lost bound to proteins filtered in urine in nephrotic syndrome)

Biopsy revealing FSGS
-Primary vrs secondary (HIV, COVID, massive obesity)
-Genetic testing if sec causes not found.