7/9/26 Morning Report with @CPSolvers

"One life, so many dreams" ***Case Presenter****: Manasa(@)* ***Case Discussants****: Rabih(@) & Lukas(@)*

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

Scribing (Krishi & Lera)

CC: 30 yo male with weakness

HPI: Reports 2 years of bl LE weakness and 8 mo of bl UE weakness.
- 2 y ago initially experienced slippage of footwear
- 6 mo agp difficulty climbing stairs and getting up from sitting
-weakness while typing on keyboard
- 4 mos ago burnt hands while cooking

ROS: no bladder / bowel Sx
Neuro: No involuntary movements, fasciculations, tremors, no numbness, paresthesia. No sleep / appetite changes.
General: No fever, LOC

PMH:
-hearing difficulty in noisy environments 1yo ago, could not be verified by test

Meds: –

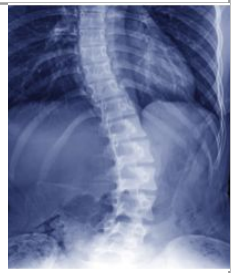
Fam Hx: —

Social Hx: No travel

Vitals: T: 38 **HR**: 80 **BP**: 140/90 lying -> 130/80 standing **RR**: 14 **BMI**: 19.2

Exam: HEENT: conscious and oriented x3
CV, **Pulm**: nl, no murmurs, vesicular breath sounds, **Abd**: normal
MSK: spine curvature, pes cavus, DIP dorsiflexion and plantar flexion of PIP all toes
Neuro: mild b/l sensorineural hearing loss localized to cochlear nerve
Strength: b.l shoulder 5/5, elbow and wrist flexors ⅘, extensors⅘; b.l hip and knee ⅘; ankle dorsi and plantar ⅘. Motor: normal bulk
Tone: hypotonia in all 4 extremities
DTR: biceps, triceps, supinator, knee, ankle reflexes absent in both limbs
Sensory: pain and temp down 30% in all 4 limbs
vibration & joint position decreased below the ankle,
b/l touch absent below elbow & knee
Cerebellar: normal, spinal convexity present Gait: high stepping

Notable Labs & Imaging:
Hematology: WBC: 8000 Hgb: 13 Plt: 380,000
Chemistry:
Cr: 0.52 BUN: 15 **Normal electrolytes**
LFTs: nl ESR:20
HIV, HBV, HCV neg
B12: 500; TSH: normal
Imaging:
EKG: nl **CXR**: nl
NCS: sensory and motor axonal neuropathy UE & LE



Gene analysis: neg for PMP22-VE, **positive** for MFN2(mitofusin 2) on chromosome 1
Dx:
CHARCOT MARIE TOOTH

Problem Representation: Young male with a progressive non inflammatory sensorimotor symmetric axonal polyneuropathy with stigma of chronicity due to pes cavus and scoliosis associated with bilateral sensorineural hearing loss

Teaching Points (Javier)

Approach
Neurologic syndrome, always to consider:
- Epidemiology
- Localization
- Compromise of upper of lower motor neuron
- Time course

Two main hypothesis
1.Specific diagnosis, 2. broad bucket etiology (What is the syndrome, what is the problem) In this case, eg. trying to solve the problem (Why is this patient coming?)
Eg. it's important to consider first cervical spine lesion.
- Although, always consider GI and GU function to rule out chronic demyelinating diseases. Do not over use it to rule out completely a medular compromise
- Sensory involvement is really useful to localize
- Proximal muscular predominancy helps localize problem in muscular etiologies.

Background features in this case MAKES consider genetic causes as Neurofibromatosis, Charcot Marie tooth disease…

Pivotal question
First principle question: What type of disease can produce these neuro- progressive symptoms? toxic substances, nutritional deficiency, genetic cause, slowly progressive compressive disease, Neurodegenerative (less probable with all the associated symptoms)
- Always to consider ALS
- Vasculitis a ddx but would be really atypical as chronic fingerprint of this etiology
- Fever can be noise, but if it persists start considering inflammatory or infectious etiologies.

Precise localization of this syndrome: symmetric bilateral polyneuropathy with motor and sensory compromise, adding the fact that this presentation is ULTRA CHRONIC→ Genetic cause, but first rule out toxic and nutrient deficiency etiologies