



12/17/24 Morning Report with @CPSolvers



"One life, so many dreams" Case Presenter: Dr. Sebastian Green (@sebfgreen) Case Discussants: Dr. Aye Thant (@AyeThant94), Dr. Aaron Berkowitz (@AaronLBerkowitz)

<p>CC: generalised weakness</p> <p>HPI: 29F right handed. Previously independent. Progressively worsening generalised weakness over 2m. Now wheelchair bound. Worsened rapidly in left leg 3d ago, then spread to right leg and arms. Difficulty swallowing since 2d. Difficulty urinating. Needed Foley catheter upon admission. Tingling in all extremities. -ve for headache, cognitive issues, etc.</p> <p>ROS: 40lb weight loss over 4 months in the context of Significant abdominal pain causing her to avoid food</p>	<p>Vitals: T: 36.9 BP: 147/110 RR: 16 HR: 125 Sat: 96%</p> <p>Exam: Gen: wasting and malnourishment HEENT: wnl CV: tachycardic, regular Abd: distended. Diffuse tenderness Neuro: <u>Mental status:</u> AOx4, Speech fluent, intact comprehension, naming, repetition. Able to spell WORLD backwards. Intact short and long term memory. <u>Cranial nerves:</u> EOMI. nystagmus disappearing after 2 beats. Hypophonic and tremulous voice. Midline uvula and tongue <u>Motor:</u> moderate wasting and atrophy symmetrically, decreased tone. Proximal 1/3 > distal 2/3 weakness bilaterally. Strength unchanged on repetition. <u>Sensory:</u> intact light touch. Impaired proprioception in toes. Can't discriminate light touch from pin prick upto shoulders and hips. <u>Reflexes:</u> absent <u>Coordination and gait:</u> could not be examined due to weakness</p>	<p>Problem Representation: 29F p/w chronic abdominal pain and 2 months of generalised weakness, and weight loss worsening progressively over the past 2 months.</p>
<p>PMH: 4m developed 2x episodes of seizure. Normal CT head. Started on Kepra.</p> <p>Chronic abdominal pain. Negative CTAP and EGDs. On chronic opioids.</p>	<p>Notable Labs & Imaging: Hematology: WBC: 7 Hgb: 12 Plt: 202k MCV: 82 Chemistry: Na: 138 K: Cr: 0.4 BUN: 8 Ca: Ph: 3.7 Mg: 1.6 Glu: 95 AST: 137 ALT: 301 ALP: 76 Bili: 1. Albumin 3.4 CK 14 TSH 0.44 Nutritional panel B12 high (taking supplements), B1 and B6 wnl, Vitamin D 18 (N>30) Infectious panel negative, Heavy metals wnl, Ganglioside panel negative ANA and ANCA negative</p> <p>Imaging: MRI brain and spine with & without contrast: nl NCS and EMG: <u>EMG:</u> fibrillation potential, sharp waves, incomplete muscle activation <u>SNAP</u> Reduced amplitude of sural, median and ulnar nerves. Normal peak <u>CMAP</u> Reduced amplitude of tibial, peroneal, median and ulnar nerves. Normal distal latency and conduction velocity</p> <p><u>Further testing:</u> urine porphobilinogen high, aminolevulinic acid (ALA) high Dx Acute intermittent porphyria complicated by axonal neuropathy <u>Clinical course:</u> patient mentioned maroon coloured urine, improved upon receiving hemin infusions. Later started givosiran for prevention. Returned to her work after rehab.</p>	<p>Teaching Points (Vale): Generalized weakness: True motor weakness vs asthenia from systemic diseases. - Corticospinal tract: Motor cortex -> corona radiata -> brain stem -> spinal cord -> roots -> nerve -> NMJ -> muscle. - Isolated generalized weakness: Muscle and NMJ are more likely. Tempo: Subacute -> inflammatory, metabolic, rapid progressive tumors. Localization: Progressive weakness + sensory + autonomic involvement = nerves > spinal cord > brainstem > b/l brain - Cervicomedullary junction compressive abnormalities can present as progressive weakness slowly affecting one limb and extending to other extremities. - Possibility of multiple lesions presenting in a young female -> MS, NMO. - Think fast: weakness + abdominal pain in a young patient -> AIP? (rare, acute onset)</p> <p>Polyneuropathy: Length-dependent vs non-length dependent - Length-dependent: Longest nerves are affected first by axonal pathology. Starts distally. Ex: DM neuropathy. - Non-length dependent: Driven by demyelination. All nerves have myelin, but the longest nerves have the most so they are usually affected most as well.</p> <p>Subacute sensorimotor demyelinating polyneuropathy Ddx: CIDP, paraprotein-associated, amyloid, autoimmune systemic disease/vasculitis, paraneoplastic, metabolic (thiamine), toxic (lead, arsenic, mercury, NO). - Nitric Oxide exposure: Induces functional B12 deficiency (dorsal column involvement)</p> <p>Axonal polyneuropathy: Metabolic and toxic causes > vasculitic neuropathy. Acute intermittent porphyria: Autosomal dominant defect in Heme metabolism. Dx by high urine porphobilinogen and ALA (Heme precursors). - SPs: Polyneuropathy (axonal and motor predominant), Pain in the abdomen (out of proportion to exam), Psychological changes, Purpuric urine, Precipitated by drugs or alcohol. - 55% acute, 18% acute on chronic. - Tx: Hemin infusions decreases heme synthesis in the liver thereby decreasing byproducts. Givosiran interfering RNA that prevents precursors build up.</p>
<p>Fam Hx: -ve</p> <p>Soc Hx: dental assistant</p> <p>Health-Related Behaviors: alcohol occasionally</p> <p>Allergies:</p>		