# 7/9/24 Morning Report with @CPSolvers

*"One life, so many dreams" **Case Presenter**:Aye Thant (@AyeThant94) **Case Discussants**: Dr.Aaron Berkowitz(@AaronLBerkowitz) and Valeria (@valeroldan23)*

**CC**: **bilateral lower limb weakness for 20 years.**

**HPI**: 67 y/o M R handed, previously well. No any preceding infection . presented after 15 years of symptoms . gradual in onset , persisting , progressive not relapsing and remitting course. Couldn't climb the stairs, stand up from a squatting position (proximal muscle weakness) no falls
or tripping at the onset .
+Muscle cramps. **Last 5 years** , gradual worsening , seek medical attention and noticed arm weakness .
+nasal voice , bilateral drooping of eyelids , **no** BV/DV, fluctuation, fatigability . nl hand grip , no SOB/dysphagia .
+ facial muscle twitching
No sensory sx , bladder or bowel sx.
**Last 3 years:** worsening of arm and leg weakness , frequent falls, decreased hand grip (distal most common affected) recent year couldn't walk , needs assistance , noticed enlargement of breast and erectile dysfunction.
Little SOB and dysphagia **now.**

**PMH**:
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**Meds**:
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**Fam Hx**:
4 siblings (M) all have bilateral weakness and ptosis
Non consanguineous marriage
Mothers siblings are all females but their sons all have weakness (every male in the family)

**Vitals: T: HR: BP: RR:** nl
**Exam: Gen:** unremarkable
**Neuro:** CN: free range of ocular movement , mild ptosis , mild weakness of orbicularis oculi ⅘ , no blurred vision . no facial drooping but there was fasciculation especially around the mouth , no uvula deviation.
Tone nl in UL but decreased in LL
Power: mostly 5
⅘ on finger abduction and extension.
Reflexed absent on knee and ankle .
Plantor : flexion
Wasting of hand muscle
Cerebellar: normal
+gynecomastia, Tongue atrophy and fasciculation.
(only motor involvement , LMN no UMNL ,chronic progressive , all four limbs with bulbar involvement and anti-androgen feature .

**Notable Labs & Imaging**:
**Chemistry**:
Anti Ach ab (- )
Ck:928 U/L
Spirometry: mild restriction PFT: nl
**Imaging**:
EKG: CXR: Echocardiogram:nl
NCS and EMG: electrophysiological evidence of wide spread very chronic denervation, partial reinnervation at many muscle from all 4 limbs and glossus muscle . consistent with chronic ant horn disease. Sensory conduction nl
**Genetic testing: analysis of CAG anti androgen receptor gene in the coding gene, showing repeat expansion (47 repeats)**
Dx: Kennedy's disease ( x-linked adult onset disease of LMN that predominantly affects limbs and bulbar muscle)

**Problem Representation**: a 67Y/O M with 20 yr hx of LL weakness, positive family hx for X-linked pattern , fasciculations, areflexia, muscle atrophy . genetic testing showing kennedy's disease .

**Teaching Points (Ethan)**:
**Bilateral LE weakness**: localization -> frontal cortex, posterior limb of pyramidal tract, brain stem, spinal cord, anterior motor neuron, NMJ, muscle
**Chronic:** consider causes like slow-growing tumor, metabolic causes (e.g., vitamin deficiency), chronic infection (e.g., HTLV-1, HIV myelopathy, Pott disease), familial causes (e.g., hereditary neuropathy), etc.
**For localization ddx:** UMN/LMN signs, distal/proximal weakness, whether sensory or autonomic deficits (would favor myelopathy with sensory/autonomic deficiencies over peripheral nerve disease process), pinpoint the myotome
**Most common cause of bilateral le weakness:** spinal cord -> muscle -> nerve. More likely to be thoracic cord or below (global peripheral process, brain lesions less likely), cauda equina, or parasagittal meningioma, bilateral ACA stroke (less likely)
Proximal LE muscle weakness + bilateral facial drooping + dysphagia (bulbar weakness) + LMN signs (fasciculation), and no sensory/autonomic deficiency -> symmetric pure motor deficits -> consider myopathy (hereditary causes with late presentation, paraneoplastic, autoimmune, etc.), ALS (not likely with this chronic), SMA, etc.
**Myopathy ddx:** inflammatory myopathies (dermatomyositis, polymyositis, inclusion body myositis), toxic (statin, steroid,, tacrolimus), genetic (myotonic dystrophy, duchenne, becker, distal muscular dystrophy, etc.), those associated with systemic disease (endocrine, autoimmune disease, etc)
**Why MG is not likely:** usually present with eye involvement, with fatigability, and fluctuation throughout the day (diurnal change). (One useful test: look at the ceiling for a while and see if the eyelids drop)
**Hypo/areflexia:** points to lower motor neuron disease (confirm with NCV/EMG)
**Elevated CK**: could also be seen in lower motor neuron disease!
**Kennedy disease (spinal and bulbar muscular atrophy):** A genetic change in the androgen receptor (AR) gene and is inherited in an X-linked recessive manner. Present with proximal and bulbar muscle weakness, gynecomastia, testicular atrophy, and infertility due to androgen insensitivity, etc. In Kennedy disease, motor neurons and spinal cord and bular system are affected, thus causing LMN signs. Currently with no tx. More indolent disease nature (compared to ALS).