LET'S TALK ABOUT...

amyotrophic lateral sclerosis

ETIOLOGY/EPIDEMIOLOGY

↳ most common Motor Neuron Disease
- 30,000 people with ALS in US
- highest incidence in Guam and Japan
- onset typically mid-late 50's
- Men > women (1.7:1)
- sporadic ALS > familial ALS
- 70-90% limb onset, 20-30% bulbar onset

↳ **risk factors:**
- disease-causing mutations (ex. SOD1, alsin)
- gender (male > female) & age
- clusters
- family hx

* 20% of hereditary cases have a defect with gene encoding copper-zinc super oxide dismutase

PROGRESSION

↳ **clinical diagnosis:**
- requires **presence** of:
  - LMN signs
  - UMN signs
  - progression of disease within a region or other regions
- requires **absence** of evidence of other diagnoses:
  - electrophysically
  - pathologically
  - through neuroimaging

↳ **disease course:** *variable!
- average duration: 27-43 months
- death within 3-5 years (usually respiratory failure)
- ↑ survival rates for: < 35-40 y/o, limb onset (vs. bulbar), and better psychological well-being

stages

EARLY	MIDDLE	LATE
• variety of abnormal signs & symptoms • minor activity limitations	• ↑ # & severity of impairments • min-mod activity limitations • participation restrictions begin to develop	• numerous & severe impairments • dependent for all activities & participation

PATHOPHYSIOLOGY

degeneration & loss of motor neurons in the spinal cord, brainstem, and brain

↳ affects anterior horn cells in spinal cord, UMN in the cortex, corticospinal tracts, and brainstem nuclei for CN V, VII, IX, X, XII

early → late

sprouting — contiguous progression — rostral/caudal

CLINICAL PRESENTATION

* highly variable →

↳ **depends on:**
- extent of motor neuron loss
- degree and combination of UMN & LMN loss
- pattern of onset and progression
- body regions affected
- stage of disease

↳ symptoms focal and asymmetrical at onset

impairments

LMN
- muscle weakness
- hyporeflexia
- hypotonicity
- atrophy
- muscle cramps
- fasciculations (muscle twitches)

UMN
- spasticity
- hyperreflexia
- clonus
- pathological reflexes (ex. Babinski)

Bulbar
- bulbar muscle weakness
- dysarthria (slurred speech)
- dysphagia (difficulty swallowing)
- sialorrhea (hypersalivation)
- pseudobulbar affect (episodes of uncontrollable laughing or crying)

PT EXAM

↳ **emphasis on:**
- psychosocial function
- respiratory function
- environmental barriers
- fatigue

↳ **take note of:**
- bulbar/respiratory impairments (refer out!)
- environmental blocks to independence
- caregiver demands & disease education
- rate of disease progression

↳ pt can use activity log [pain level, fatigue, effort]

↳ **evaluation:**
- use to develop goals: disease progression, specific impairments, phase of disease, & activity limitations

↳ plan of care:
- goal development
  - progressive diagnosis
  - will NOT cure or delay progression
- broad goals to maximize independence & have + QOL
- address psychosocial & caregiver issues

outcome measures

ALS Functional Rating Scale (ALSFRS & ALSFRS-R):
- ICF: Activity
- measures functional status of ALS patients
- 0-4 scale:
  - → 0: unable to attempt the task
  - → 4: normal function
- reliable & valid for measuring decline in function related to muscle weakness
- revised version has respiratory items (ALSFRS-R)
- can indicate what to prioritize and if referral is necessary

ALS Assessment Questionnaire (ALSAQ-40)
- 40 item questionnaire specific to QOL
- 5 areas of health:
  - mobility
  - ADL
  - eating/drinking
  - communication
  - emotional functioning
- 0-4 scale: worst → best health status
- revised version has 11 items & is valid & reliable

CLINICAL MANAGEMENT

→ **MEDICAL MANAGEMENT:**
- No cure
- often palliative treatment
- symptomatic management
- medication (ex. Riluzole)
- cannabis (for spasms/spasticity)

* improved lifespan w/ multidisciplinary approach!

→ **PHYSICAL THERAPY GOALS:**
- maintain mobility & independence
- maintain strength & endurance
- prevent secondary complications
- management of energy consumption & respiratory fxn
- determine necessary adaptive equipment
- eliminate or prevent pain

physical therapy interventions

Exercise & ALS * use your CLINICAL JUDGEMENT
- moderately intense exercise may slow progression of hereditary forms, including copper-zinc superoxide dismutase
- mod intensity exercise has been found to ↑ QM scores, strength, functional independence, & respiratory fxn

Negative Outcomes of Exercise
- **overwork damage:** collateral sprouting could be impaired in denervated muscles w/ < 1/3 motor units
- **damage to normal muscle fibers w/ repetitive eccentrics:** may not have repair capabilities
- **exercise intolerance:** mitochondrial dysfunction, abnormal muscle metabolism, impaired activation, central activation failure → fatigue

* need constant monitoring of fatigue & weakness

Exercise Prescriptions
- manual resistance exercise using PNF:
  - ↳ during early stages, for muscle grades 4-5/5
  - ↳ generally no bad effects
- supervised gentle exercises
  - ↳ decreased pain/stiffness & improved psychosocial
- aerobic endurance
  - ↳ 10-15 min (↓ due to fatigue or ↓ motor units)
- formal & enjoyable physical activity
- strengthening (concentrics, mod resistance, grades >3+/5, monitor for overwork weakness
- endurance (monitor fatigue, < 15 min, rest)
- monitor fluid & oxygenation
- supportive, adaptive, patient & family focused

ALS STAGING & REHAB

PHASE I: INDEPENDENT

Stage 1

Characteristics
- mildly weak
- clumsiness
- ambulatory
- independent w/ ADL's

Treatment
- continue normal activity or increase if sedentary
- gentle PRE strengthening & stretching
- psych referral if needed

Stage 2

Characteristics
- mod-selective weakness
- ambulatory
- slight ↓ in ADL independence (climbing stairs, dressing)

Treatment
- stretching
- cautious strengthening for MMT 3+/5
- orthotic and AD

Stage 3

Characteristics
- severe weakness (ankles, wrists, hands)
- increase in respiratory effort

Treatment
- continue stage 2 program
- monitor fatigue
- maintain physical independence
- breathing exercises, chest stretching, postural drainage, w/c prescription

PHASE II: PARTIALLY INDEPENDENT

Stage 4

Characteristics
- Hanging Arm Syndrome
- shoulder pain w/ edematous hand
- w/c dependent
- severe LE weakness
- can perform ADL's with fatigue

Treatment
- Heat/massage for spasm
- Edema prevention
- AA & PROM of limbs
- isometric contractions to tolerance
- orthotic UE support (slings/trays
- power mobility

Stage 5

Characteristics
- severe LE weakness
- mod-severe UE weakness
- w/c dependent
- dependent with ADL's
- skin breakdown

Treatment
- family training for transfers
- positioning to ↓ skin breakdown
- turning schedule
- home modifications for mobility
- hospital bed with air mattress
- possible home mechanical vent (HMV)

PHASE III: DEPENDENT

Stage 6

Characteristics
- bedridden
- completely dependent in ADL's

Treatment
- continue stage 5 program
- impairment specific:
  - ↳ dysphagia: soft diet, adaptive feeding equipment, tube feed
  - ↳ dysarthria: speech amplification, palatal lift
  - ↳ respiratory: clear airway, trach, vent, meds to control dyspnea

LET'S TALK ABOUT...

huntington's disease

ETIOLOGY/EPIDEMIOLOGY

↳ typical onset: 40-50 y/o
↳ prevalence ~5-10/100,000 persons [rare]
↳ men > women
↳ genetic mutation:
- abnormal folding of proteins in the neurological structures of the brain & basal ganglia
- >40 "CAG" repeats on HTT allele of chromosome 4
- leads to neuro impairment → cortical destruction

↳ late stage profound dementia
↳ may have mood/psychiatric involvement
↳ life expectancy: 15-25 years beyond symptom onset
↳ progressive degeneration & atrophy of basal ganglia

PROGRESSION

↳ clinical diagnosis based on:
- onset of symptoms and family hx of HD
- genetic testing to confirm HTT allele & CAG repeats
- neuroimaging to identify changing in basal ganglia & cortical atrophy

↳ presentation changes w/ disease progression:
- early = hyperkinetics
- late = hypokinetics

* disease progression cannot be altered in rehab

stages

EARLY	MIDDLE	LATE
• variety of abnormal signs & symptoms • minor activity limitations • variable participation restrictions	• ↑ # & severity of impairments • min-mod activity limitations • participation restrictions more pronounced	• numerous & severe impairments • dependent for all activities & participation

MEDICAL MANAGEMENT

- CNS depressants to control chorea
  - Klonopin, Haldol, or Tetrabenzine
- Multidisciplinary approach
  - OT, SLP, social work, psych, MD, DO, RN
- Symptomatic management
  - Rx for tone, palliative care in late stages

PATHOPHYSIOLOGY

autosomal dominant inherited hyperkinetic movement disorder

↳ "Hereditary cortical neurodegeneration"
↳ disease of basal ganglia structures
↳ hallmark signs:
- choreiform movements
- late stage profound dementia w/ cortical destruction
- atrophy of the caudate nucleus & enlarged ventricles

CLINICAL PRESENTATION

↳ severity may be variable → depends on extent of atrophy, # of deleterious CAG repeats, & rate of progression

impairments

↳ choreiform hyperkinetic movement:
- rapid, jerky, writhing, uncomfortable
- involuntary movement at rest & with movement
- late stages: "mixed" movement disorder w/
- elements of hypokinetics due to destruction to basal ganglia structures (ex. freezing, shuffling gait)

↳ typically see UMN signs & symptoms:
- muscle weakness, fatigue, and atrophy
- hyperreflexia
- difficulty swallowing & communicating
- hypertonicity
- poor balance & trunk control → falls
- impaired mood & cognition

PHYSICAL THERAPY

↳ NMD commonly used functional measurements:
- 10m walk
- 4-stair climb
- time to rise from floor
- TUG
- 30 sec chair stand test
- 2 or 6MWT
- Brooke/Vignos scale
- GMFM

↳ OM: Unified Huntington's Disease Rating Scale
↳ rehab management: * monitor fatigue
- rhythm & cadence (auditory)
- smaller amp. motor control
- relaxation techniques
- PNF & rhythmic stabilization
- therapy after meds
- stretching
- splints/casting
- AD/DME
- TENS
- family education

LET'S TALK ABOUT...

cerebellar ataxia

ETIOLOGY/EPIDEMIOLOGY

Causes

Acquired
- stroke (<5% all)
- tumor
- structural (chiari malformation)
- toxicity
- immune-mediated (MS)
- Trauma
- infection
- endocrine

Degenerative

Non-hereditary
- multiple systems atrophy (MSA)
- idiopathic late-onset cerebellar ataxia
- immune-mediated (MS)

Hereditary
- autosomal dominant disorders (SCA)
- autosomal recessive disorders
- X-linked disorders

* SCA6: most common of SCA

cerebellar brain tumor:
- children > adults
- common location: posterior fossa
- poorer prognosis in adults
- damage of deep cerebellar nuclei can predict recovery better than age

spinocerebellar Ataxias (SCA):
- 30 known distinct SCAs - named by #
- onset is mid-later life
- genetic counseling needed before testing (hereditary)
- no pharm interventions (meds)

Episodic Ataxia:
- brief bouts of ataxia usually due to excitement, stress, or exercise
- may respond to meds
- minutes to hours episodes

STROKE PRESENTATION

cerebellar stroke

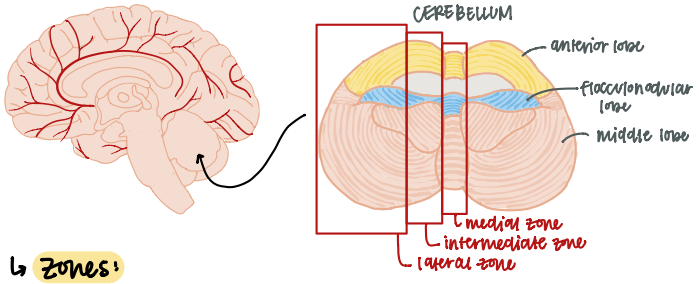
* recovery is best when deep cerebellar nuclei are not involved

↳ **superior cerebellar A:**
- dysmetria of ipsilateral arm, unsteady walking, dysarthria, nystagmus

↳ **Anterior inferior cerebellar A:**
- dysmetria, vestibular signs, facial sensory loss

↳ **posterior inferior cerebellar A:**
- vertigo, unsteadiness, walking ataxia, nystagmus

PATHOPHYSIOLOGY



↳ **Zones:**
- **medial zone:** afferent sensorimotor state of limbs
  - motor output of posture, muscle tone, upright stance, locomotion & in gaze of eyes
- **intermediate zone:** controls coordination of agonist-antagonist muscle pairs during movement (walking, limb control)
  * medial + intermediate = spinocerebellum
- **lateral zone:** controls complex, multi-joint voluntary limb movement, especially with visual guidance
  - planning of complex movements and error detection
- **flocculonodular lobe (4th zone):** helps control eye movement and balance
  - responsible for the health of the vestibulocular reflex (VOR)

↳ **Cerebellar function:**
- responsible for coordination & adaptation of movement, vestibular, balance, & motor learning
- connections to non-motor regions (pre-frontal cortex)
- can impact cognition

IMPAIRMENTS

motor
- ataxia (1° impairment)
- dysmetria
- dyssynergia
- dysdiadochokinesia
- decompensation (reduce ataxia)
- rebound affect (isometric & release)
- action tremor
- hypotonia
- imbalance in gait
- oculomotor/speech issues
- motor learning deficits

non-motor
- language processing
- working memory
- learning associations
- higher order executive fxn
- loss of control over emotional behaviors

PT EXAM

↳ **determine if disease is progressive or non-progressive:**
- if progressive → health promotion approach
- if non-progressive → recovery/restorative approach

↳ **ataxic gait presentation should be present:**
- imbalanced, slow
- shortened steps with uneven step lengths
- irregular timing → abnormal trajectory of limb
- wide BOS
- overly high step (excessive knee & hip flexion)
- uncontrolled lowering of foot in stance phase of gait
- veering along path & difficulty w/ turning & stop

↳ **systems review:**
- fatigability → increased effort of movement
- can test CV & MSK endurance

↳ **tests & measures:**
- muscle tone → hypotonia
- voluntary movement coordination
- static/dynamic balance
- oculomotor performance → smooth pursuit, saccades, gaze evoked nystagmus
- look at gait & transfers

↳ **non-progressive ataxia prognosis:**
- 1st time ischemic stroke → excellent
- 93% minimal to no residual deficits

↳ **progressive ataxia prognosis:**
- progressively worsening

outcome measures

International Cooperative Ataxia Rating Scale (ICARES)
- ICF: activity
- 19 tasks, 4 categories
  - ↳ postural control, limb movements, speech, & oculomotor
- ordinal scale, scored 0-100
- reliable & valid in progressive & non-progressive ataxias

Scale for Assessment and Rating of Ataxia (SARA)
- ICF: Activity
- quantifies performance, not categorized
- 8 items (similar to items on ICARES)
- ordinal scale, scored 0-40
- reliable and valid for SCA's

PT MANAGEMENT

↳ **lack of intervention evidence:**
- NO RCT's for interventions
- most effective intervention hasn't been established
- non-randomized, non-controlled, small sample size literature are the only ones available

evidence suggests

Gait and balance
- part task → kneeling, sitting, quadruped
- gaze, static stance, dynamic, complex gait activities
- address specific impairments if affects outcome
  ex) ankle mobility
- locomotor training with body weight support
- treadmill training
- visual guided stepping

*interventions must be challenging & be trial & error

Aerobic Exercise & Resistance Training
- for those who will not return to baseline
- can reduce fatigability (may reduce falls)

Intensity & Longer Duration
- partial relearning is possible - 10 hrs/wk, 6 months

Compensatory strategies
- slow down movement
- decomposition
- visual cues
- minimize distractions
- widen gait
- use AD only if safely used

Biofeedback/EMG
- auditory feedback for stepping speed & length
- biofeedback + relaxation techniques to ↓ tremors & improve feeling
- visual biofeedback of COP for postural sway

Brain Stimulation
- ↓ motor cortex excitability
- stimulation of cerebellum → ↑ in balance, ataxia, & gait

Weights
- slows movement, creates difficulty at distal limb
- no carryover → no benefit

Reducing UE tremor & ataxia
- manipulation of visual info can improve movement

Lycra compression garments
- insufficient evidence

Promote mobility independence
- use power mobility to promote independence

SUGGESTED TREATMENT

impairment	treatment
Poor coordination & grading of muscle power	Reduce degrees of freedom, external device, postural control
Reduced adaptability in environment	Graded exposure, sensory cues, conscious attention
Reduced automaticity of walking	Stepwise prompts, high reps, conscious attention
Reduced postural control, tone, & tremor	High intensity and conscious attention
Altered timing of stepping	Specific strength & balance, compensation, environmental cues, aids Avoid dual task, conscious attention to gait

types of ataxia	treatment
Spinocerebellum involvement	Strength/balance, walking aides, cortically control walking, cues
Vestibulocerebellum involvement	Compensatory head fixing or retain VOR, strength/balance
Cerebrocerebellum involvement	Retrain gait in all functional environments, avoid obstacles