Considerations for the Plan of Care for Patients with Huntington's Disease

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Objectives

- Be able to articulate the disease process of HD.
- List several impairments that are likely to be observed when evaluating a patient with HD.
- Accurately identify the stages of HD and conceptualize how they relate to the treatment progression.
- Incorporate exercise guidelines to improve independence, decrease fall risk, and improve quality of life.
- Identify communication and mental health considerations in the plan of care for patients with HD.
- Apply plan of care considerations to a case study model to simulate critical thinking in a patient-care scenario.

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Huntington's Disease Overview

- Neurodegenerative disease
- HD etiology[1]
  - Autosomal dominant inherited CAG trinucleotide repeat expansion in HTT gene on chromosome 4
    - Production of mutant huntingtin protein (mHTT)
    - >39 CAG repeats - disease develops
    - 36-39 CAG repeats - reduced penetrance

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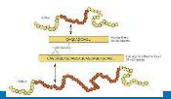
Huntington's Disease Overview

- HD Epidemiology[1]
  - Prevalence: ~13.7 per 100,000 people in Western populations
  - Incidence: 4.7-6.9 new cases per 1 million per year



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Huntington's Disease Pathogenesis[1]

- Toxic mutant huntingtin protein (mHTT)
- Medium spiny neurons (MSN) of the striatum
  - Especially vulnerable to the effects of mHTT
- Striatal pathology
  - Biphasic pathology
- Post-mortem studies
  - Diffuse atrophy of caudate and putamen

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Huntington's Disease Diagnosis[1]

- Median age of diagnosis: 40 years old
- Men and women affected equally
- Diagnostic process:
  - Family history
  - Positive genetic test/onset of motor symptoms
    - Unified HD Rating Scale - Total Motor Score
- Psychiatric symptoms precede motor symptoms

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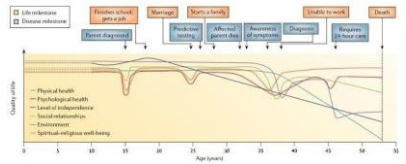

HD - Multidisciplinary Approach

"The complex nature of Huntington's disease makes it unlikely that any one professional will have all the skills needed to help any one individual. It is therefore of utmost importance that the service providers take a multidisciplinary approach to Huntington's disease in order to identify the best way to assist individual patients by taking into account their differing needs."

Benefits of Exercise

- One study showed that in mice with HD in an enriched setting[2]:
  - Motor function preserved
  - Delayed symptom onset
- With respect to humans, several studies support exercise in people with HD[3]

Huntington's Disease Stages

1. Pre-Symptomatic /Early Stage
2. Middle Stage
3. Late Stage

Clinical stages of Huntington's disease (HD)

Early	Middle	Late
Clumsiness	Chorea (mild)	Chorea (moderate-severe)
Apathy	Dystonia	Parkinsonism
Anxiety	Balance and gait difficulties	Dysphagia
Agitation	In-coordination	Dysarthria
Irritability	Weight loss	Self-neglect
Personality changes	Disinhibition	Dementia
–	Cognitive impairment	Hallucinations/delusions

Ref: Modified from http://www.ncbi.nlm.nih.gov/books/NBK1009/

Huntington's Disease - Early Stage[4]

- Main impairments:
  - Fine motor deficits
  - Chorea
  - Balance and flexibility impairments
  - Unsteady gait pattern

Early Stage Exercise Considerations[4]

- Aerobic exercise
  - Walking (treadmill/overground)
  - Stationary bike
  - Swimming
- Strengthening (focus on postural muscles)
- ROM/stretching
- Coordination activities
- Dynamic/static balance interventions

Huntington's Disease - Middle Stage[4]

Main Impairments:

- Dystonia
- Chorea
- ROM restrictions
- Stabilizer muscle weakness
- Increased balance/gait deficits (falls)

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Huntington's Disease - Late Stage[4]

- Main Impairments:
  - Postural changes
  - Respiratory limitations (pneumonia risk)
  - Mobility impairments and falls

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Middle-Late Stage Exercise Considerations[4]

- Shorter exercise duration
- Make the exercise functional
  - Patient-centered care
  - Ex: transfers, hand exercises, sit to stand, walking

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Neuropsychiatric Considerations

- Neuropsychiatric symptoms can occur **decades** before the motor symptoms[5]
  - Psychosis
  - Apathy
  - Depression & anxiety
  - Irritability/aggression
  - Obsessive compulsive behaviors
  - Sleep disorders
  - Cognitive dysfunction
  - Suicidal ideation

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Systematic Review Study - HD[6]

- Six studies included
- Studies implemented structured endurance and/or resistance training programs
  - Self-selected gait speed improved
  - Berg Balance Scale improved
  - No significant/stable cognitive variables
  - Increase in predicted VO2max
  - Mental component of SF-36 improved

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Clinical Practice Guideline Recommendations - PT[7]

- Mixed-Methods Systematic Review
  - 23 quantitative studies
  - 3 qualitative studies
- Upcoming RCT in an longitudinal observational study*
  - *Results not yet published*

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Clinical Practice Guideline Recommendations - PT[7]

- Grade A Evidence
  - Aerobic Exercise (+/- resistance training):
    - Improvement in fitness and motor function
  - Supervised Gait Training:
    - Improvement in spatiotemporal features of gait

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Clinical Practice Guideline Recommendations - PT[7]

- Grade B Evidence
  - Exercise training improves balance
  - Inspiratory and expiratory training improves breathing function and capacity
  - Transfer training and caregiver education regarding physical activity during mid-stages of HD may improve performance

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Clinical Practice Guideline Recommendations - PT[7]

- Expert Consensus (late-stage HD)
  - Positioning devices
  - Seating adaptations
  - Caregiver training

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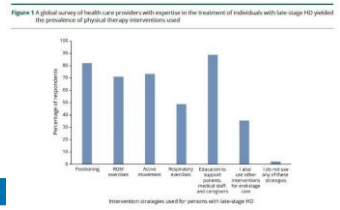
Late-Stage HD - Global Survey of PT Interventions[7]



Action Statement #1 - Aerobic Exercise[7]

- Moderate Intensity (55-90% HRM)
- 3x/week, minimum of 12 weeks
- **Recommendation strength: strong**

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Action Statement #2: Gait Training[7]

- One-on-one, supervised gait training
  - Improves spatiotemporal measures
  - Ex: walking speed, step length
- **Recommendation strength: strong**

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Action Statement #3: Balance Exercises[7]

- Individualized balance exercises
- Variety of intervention durations and frequency
- Recommendation strength: weak

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Action Statement #4: Breathing Exercises[7]

- Effectiveness of breathing exercises in persons with HD
- Recommendation strength: weak

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Action Statement #5: Postural Control[7]

- Effectiveness of postural control training in persons with HD
- Recommendation strength: weak
- Expert consensus: intervention for MSK and postural changes should be tailored to the stage of the disease.

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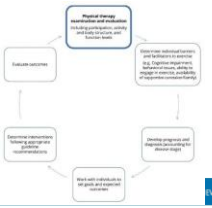
Broda Chair

Action Statement #6: Late-Stage Care[7]

- Role of PT on ADLs, seating and positioning in late-stage care
- Recommendation strength: expert opinion
  - Establish realistic goals
  - Enhance patient quality of life
  - Decrease caregiver burden

Patient Client Management - HD[7]



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Overall CPG Recommendations[7]

- Perceived benefits
- Shared goal setting - caregiver and patient
- Benefit of exercise interventions multiple times over a year or longer
- Physical activity -> secondary impairment prevention
- Individualized exercise plan with involvement of a caregiver

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PAT-HD Tool[8]

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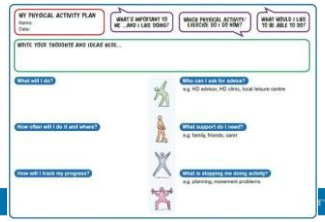
PAT-HD Tool[8]



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Unified Huntington's Disease Rating Scale[9]

- Part I: Motor Function
  - Higher scores: inability to complete motor task
- Part II: Cognitive Function
  - Higher scores: better cognitive performance
- Part III: Behavioral Assessment
  - Higher scores: severe behavioral symptoms
- Part IV: Functional Capacity
  - Higher scores: higher functional status

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Fall Prevention in HD[4]

- Falls typically start to occur in the middle stages
- Common catalysts for falls:
  - Navigation of stairs
  - Multi-tasking
  - Fast turns while holding something
  - Stepping over things on the floor
  - Shoes with poor support

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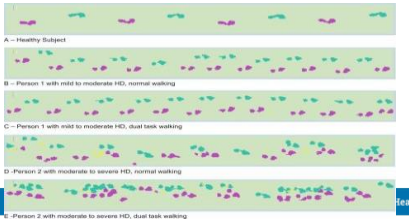
What impairments lead to increased fall risk in HD?[4]

- Balance impairments
  - Increased sway in standing
  - Delayed balance reactions
  - Increased challenge with tandem stance/walking
- Gait impairments
  - Decreased walking speed
  - Short steps lengths
  - Wide base of support
  - Veering path
  - Uneven step length

Assistive Device Recommendations-HD

Gait Mat Data - HD[10]



Gait training considerations in HD[5, 11]

- Rhythmic haptic cueing (RHC)
  - Significant improvement in gait coordination
- Ambulating to a metronome
  - Improved gait speed
- Treadmill training

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Balance Considerations in HD[5]

- Treatment Interventions:
  - Firm surface, compliant surface, eyes open/closed
  - Stepping reactions
  - Safe setup, involve the caregiver
  - Wii and Dance Dance Revolution

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Additional Fall Risk Considerations in HD[5]

- Bedroom safety
  - Bed rails
  - Lower bed height
  - Enclosure bed
  - Craig bed

- Clothing/devices:
  - Velcro, high top shoes
  - Soft helmets, knee/elbow pads, hip protector pads

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Speech Therapy Outcomes Measures[12]

- Patient reported outcomes
  - HDQLIFE Speech Difficulties
  - HDQLIFE Swallowing Difficulties
  - Communication Participation Item Bank (CPIB)

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Communication Impairments - HD[13]

- Hyperkinetic dysarthria
- Cognitive-communication impairments
  - Predictable/simple phrases
  - Decreased initiation
  - Topic maintenance difficulty
  - Shifting during conversation
  - Perseverative behavior

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Augmentative and Alternative Communication - HD[13]

- Stage 1 (no detectable speech disorder)
  - Use of memory and organization aids
- Stage 2 (speech disorder, but intelligible)
  - Continued use of memory aids, behavioral strategies to improve success with face-to-face communication
- Stage 3 (speech intelligibility is reduced)
  - Ex: cueing board, alphabet or word board with large spaces, orienting cues, partner training)

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Augmentative and Alternative Communication - HD Cont. [13]

- Stage 4 (Natural speech must be supplemented by AAC)
  - Previous strategies as well as low-technology direct selection systems
- Stage 5 (Speech is not functional)
  - Simple devices with common requests/words, a yes/no system, and communication partner training are recommended

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Augmentative and Alternative Communication - HD Cont. [13]

- Dystonia, rigidity, and chorea are all factors to consider when choosing communication strategies
- A communication app on a tablet can be beneficial and personalized
- Customized communication books are options when technological options (i.e. AAC with dynamic display screen and eye gaze access) are too expensive

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Case Study

You are chart reviewing a patient for an outpatient physical therapy evaluation. The patient is a 37 y.o. woman with Huntington's Disease. She presents to physical therapy with the main complaint of an increase in the number of falls she is having at home. When you perform your handoff with speech therapy, they tell you that the patient has mild cognitive impairment. Upon evaluation, you discern that the patient has weakness in the postural musculature, dystonia, chorea, and decreased overall ROM in all four extremities. The patient scores a 49/56 on the Berg and a 20/30 on the FGA. The patient's partner is worried that exercise may make the patient's impairments worse and lead to increased disease progression.

Case Study - Discussion

1. What stage in the HD process is this patient likely in based on patient presentation?
2. What treatment interventions would you like to begin implementing into your plan of care based on impairments?
3. What caregiver education can you provide at the evaluation? Throughout the plan of care?
4. What screening questions could you ask at evaluation to see if additional referrals are required?
5. What are specific aspects of gait training that you would include for this patient from the HD research discussed?

Questions?

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