Living with Early HD

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Living with Early HD

Outline

- Definitions of early HD
- Trajectory: from pre-HD to HD
- Scenarios to consider
- Strategies
  - Small changes can make a difference
HD Over the Lifetime

Neurobiological marker (arbitrary units)

Age

Diagnostic (motor) threshold

CAG < 30
CAG > 39 Untreated

Paulsen JS, Hayden M, Stout JC and the PREDICT-HD Investigators.
Huntington Disease Stages: Functional rating scale

**Stage 0:** Pre-HD

**Stage 1:** Slightly lower performance at work; independent at home

**Stage 2:** Can still work (lower level), still mostly independent at home

**Stage 3:** Difficult to work, starts to needs help with financial, home activities

**Stage 4:** Unable to work. Needs major assistance with care

**Stage 5:** Full-time nursing care care required

*Adapted from Shoulson et al, Quantification of Neurological Deficit, Boston: Butterworth, 1989*
# Total Functional Capacity (TFC) Score

## Abilities to:

<table>
<thead>
<tr>
<th>Abilities to:</th>
<th>Points</th>
</tr>
</thead>
<tbody>
<tr>
<td>Work</td>
<td>0 - 3</td>
</tr>
<tr>
<td>Handle finances</td>
<td>0 - 2</td>
</tr>
<tr>
<td>Do home activities</td>
<td>0 - 3</td>
</tr>
<tr>
<td>Perform self-care</td>
<td>0 - 3</td>
</tr>
<tr>
<td>Live at home</td>
<td>0 - 2</td>
</tr>
</tbody>
</table>

**TOTAL SCORE** 0 - 13

## HD Staging

<table>
<thead>
<tr>
<th>HD Staging</th>
<th>Points</th>
</tr>
</thead>
<tbody>
<tr>
<td>Stage 1</td>
<td>11 - 13</td>
</tr>
<tr>
<td>Stage 2</td>
<td>7 - 10</td>
</tr>
<tr>
<td>Stage 3</td>
<td>4 - 6</td>
</tr>
<tr>
<td>Stage 4</td>
<td>1 - 3</td>
</tr>
<tr>
<td>Stage 5</td>
<td>0</td>
</tr>
</tbody>
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TFC declines by 0.7 units/year

Marder et al, *Neurology* 2000;54;452-8
Symptoms in Huntington’s disease

MRI views of Striatum

- Impulsivity
- Balance Problems
- Creativity
- Slow Eye Movements
- Episodic Anger, Irritability
- Slowness of Movement
- Depression, Anxiety
- Trouble Swallowing
- OCD Psychosis
- Chorea: Involuntary Movements
- Multi-tasking
- Restless, Fidgets
- Organizing Concentrating Prioritizing
- Fine Motor Tasks

Aylward EH. Brain Res Bull 2007;72:152-8
The Transition from Pre-HD to HD

Current clinical definition: Clear presence of a movement disorder.

But.......brain changes and other symptoms start years before.
Studies in Pre-HD: Predict-HD

- **WHEN:** 2000 – current
- **WHERE:** >20 sites in US, Canada, Australia, Europe
- **WHO:** Participants who know their CAG repeat status, CAG expanded +/-, not diagnosed
- **WHAT:** MRI brain scans, tests of thinking, tests of movement, questions about symptoms
- **Findings:** MRI changes, performance on thinking tests, tests of movement and sensation begin to change at least 10 years before diagnosis

At the time of definitive diagnosis of HD, the striatum has already lost 50% of its volume.

Studies in Pre-HD: TRACK-HD

- **WHEN:** 2008
- **WHERE:** 5 sites in Europe, Canada and US
- **WHO:** 336 (People with pre-HD, early HD and controls)
- **WHAT:** Scans, motor, thinking tests
- **Findings**

Early HD encompasses the period when symptoms first began to appear, through the time of initial diagnosis into Stages 1 and 2.

- Symptoms:
  - Functional changes at work and home
  - Cognitive changes
  - Mood and behavior changes
- Quality of life begins to change
- Caregiving begins before diagnosis
The goals of living with early HD involve maintaining all of the following for both the persons with HD and their families:

- Autonomy
- Dignity
- Meaningful social interaction
- Communication
- Comfort
- Safety and order
- Spirituality
- Enjoyment, entertainment and well-being
- Nutrition
Managing the Trajectory

Don’t wait for the “official “diagnosis –

1. Establish care with an HD specialist if changes are beginning to appear.

2. Access reliable information about HD from recognized advocacy organizations and websites.

3. Take good care of your brain: healthy diet, regular exercise, avoid smoking and excessive alcohol use.

4. Consider volunteering for research: observational studies or clinical trials.

Source: T. Tempkin RNC, MSN, ANP
What about medications and supplements to delay onset or slow progression of HD?

- **Minocycline**: no evidence for benefit
- **Coenzyme Q10**:
  - studies suggest possible benefit
  - demonstrated tolerability in people with the HD gene expansion
  - high dose needed, $$$
  - large multicenter trial now underway (2CARE, Cudkowicz, PI), enrollment closed
  - Pre-HD study completed (PREQUEL, Ross, PI); results will be announced soon
- **Creatine**:
  - studies suggest possible benefit
  - good tolerability in people with the HD gene expansion
  - but high doses can be toxic!
  - Large multicenter trial underway (CREST-E, Hersch PI)
- **Fish Oil (ethyl EPA)**:
  - not helpful for chorea
  - Good tolerability in people with the HD gene expansion
  - ?Other benefits?
Scenario 1

A 38 year old woman who tested positive for the HD CAG expansion 5 years ago comes to the clinic to establish care. She has never experienced chorea or clumsiness, but she has had persistent feelings of sadness and hopelessness, sleeps poorly, and has lost weight. She moved from one job to another in the 2 years, and gives vague reasons for the job changes. She admits that she’s missed paying some bills on time, and that she doesn’t keep her apartment as clean as she used to. She wonders if she might be starting to get symptoms of HD.

What do you think?
Managing the Trajectory: Stage I

Changes
- Still working— but may be stressed or starting to decline in performance
- Still independent at home, but multi-tasking begins to become more difficult
- Emotional symptoms of anxiety, depression, irritability may occur

Strategies
- Stress management
- Adaptations at work
- Allow more time; avoid multi-tasking
- Seek treatment for cognitive and mood changes
- Counseling
- Medications
Scenario 2

A 28 year old man with the HD CAG expansion gets angry at work and assaults a co-worker. He quits that job and is re-hired at another, and no legal problems have surfaced. He denies difficulty with irritability, but his wife reports frequent anger outbursts at home. His neurological exam shows only questionable signs of HD.

What do you think?
Managing irritability and anger

- Establish and stick to routines, schedules
- Look for behavioral triggers; re-direct
- Avoid confrontations and ultimatums
- Limit alcohol
- Limit stimulants (caffeine)
- Consider anxiety or depression as a cause
- If anger is severe and/or frequent, medications will help
- Call authorities if necessary

Scenario 3

A 25 year old woman is brought to the HD clinic by her father. Her mother died of HD in her 30’s, and her older sister was diagnosed with HD a few years ago. She’s had difficulty keeping a job for the last year or so, and she is more withdrawn and apathetic. Her neurological exam shows that she has mild memory problems, marked slowness in movement, rigidity in her arms, poor balance.

What do you think?
Managing the Trajectory: Stage II

Changes
- May still be able to work – but at a reduced capacity.
- Still mostly independent at home, but organizing and prioritizing may be more challenging, and may not be as engaged in family activities.

Strategies
- If possible, negotiate with workplace for modified duties, or apply for disability
- Help to maintain a schedule
- Seek treatment for cognitive and mood changes
- Future planning
  - Disability
  - Long-term care
Managing Apathy

- Routines, structure, “prompting”
- Evaluate for Depression
- Medical issues (thyroid dysfunction)
- Side effects of medications, including those for chorea, insomnia, allergies, depression
- Trial of atomoxetine failed to show benefit
- Sometimes stimulant drugs can be remarkably effective

Scenario 4

A 29 year old successful young professional developed psychosis as the initial manifestation of HD. His psychosis is well controlled with medications. By age 36 he can no longer work, but externalizes the reason. His wife divorces him, he no longer has a car, and he comes to clinic expressly to “get my car keys back.” He insists that he doesn’t have HD.

What do you think?
Unawareness and HD

- Not the same as psychological denial
- It’s quite common, but not everyone with HD has this difficulty
- Can be limited to some symptoms, but not all
- Can lead to work, family conflict, injury
- Treatment is difficult; behavioral strategies are best

A Few Final Words

- It’s never too early to start behavioral strategies
  - Structure and routines help tremendously
  - Regular schedule of sleep/wake, meals, activities
  - Use calendars and reminders
  - The HD mantra: one thing at a time

- Good for the brain:
  - Healthy diet
  - Regular exercise
  - Avoid brain toxins…..alcohol, smoking

- Caregiving begins BEFORE Stage 1.
  - Ask for help.
  - “Share the Care!”
Thank you, patients and families!

Thank you for your courage and hope for the future … for your advocacy for better care and a cure … for teaching health professionals about HD … and for volunteering for research.