



PALLIATIVE CARE CASE OF THE MONTH

“Huntington's Disease at End of Life” Caitlin Siropaides, MD

Volume 16, No. 60

April 2016

Case: Ms. P was a 44-year-old female with history of Huntington's disease diagnosed at age 30 who presented with worsening agitation, combativeness, impulsivity, and uncontrolled chorea. Her hospitalization was complicated by recurrent aspiration pneumonia and injuries related to falls. The palliative care team was consulted to discuss goals of care with the Power of Attorney four months into her admission. The team was concerned that despite their conversations, the POA's wishes for a focus for quality of life was not consistent with the treatments she was receiving.

History of Present Illness: She was unable to ambulate and would lunge herself forward when trying to stand. She was, therefore, isolated in an empty padded room with mattresses covering the ground. She was unable to feed herself and would aspirate frequently due to impulsivity with feedings. Her weight was low but appeared stable. She was usually mute, but occasionally would grunt or scream randomly. She did not interact or participate with staff, but was noted to interact with a childhood friend. In order to minimize injuries and combativeness, she required a complex regimen of anti-psychotics and sedatives managed by psychiatry.

Social History: The patient had estranged herself from all family, including her only daughter, at the time of her diagnosis due to fear of them witnessing the disease progression. The patient had observed her mother die from the disease, though they were not aware of the diagnosis at that time. Her only support was her POA, a close childhood friend.

Discussion: Huntington's disease is a hereditary neurodegenerative disorder caused by an expansion of a repeating CAG triplet on chromosome 4 which results in a protein with an abnormally long polyglutamine sequence. This abnormal protein is toxic to brain cells, particularly the corpus striatum of the basal ganglia, and involves other regions of the brain in later stages. The disease manifests as a triad of motor, cognitive, and psychiatric symptoms. Its trajectory varies considerably amongst the affected population with an average length of survival after diagnosis of 10-20 years. The movement disorder includes chorea, which is usually progressive and can become very severe in later stages, or may be replaced by rigidity, dystonia, and bradykinesia which affects balance resulting in falls.

The motor abnormality can also cause impairment of speech and swallowing leading to aspiration. The cognitive component consists of a slowing of mental processing and dementia in late stages. Though memory, language and conceptual abilities remain intact, a reduction of speech results in limited communication. The psychiatric component of the disease has a wide range of severity, though most develop some level of personality and behavioral changes including apathy, irritability, impulsivity or obsessive/compulsive traits.¹

Advance care planning is an important component of patient care as patients lose the ability to communicate complex or abstract thoughts in late stages. Patients should be prompted to designate a POA early in their disease and discuss end-of-life preferences. This should include plans for location of care as most HD patients with advanced disease are unable to remain at home, and many nursing homes are unwilling or not equipped to care for patients with impulsive or explosive behaviors. The Huntington's Disease Society of America Center of Excellence is a network of support to provide information and assistance to families and providers (www.hdsa.org). Their specialized social workers are often familiar with facilities that are able to accommodate this patient population.

Other issues of importance for advance care planning include patient preferences regarding artificial nutrition and hydration. In some patients, swallowing impairment can occur early in the disease course while functional status and quality of life are relatively preserved. In these cases, it may be reasonable to consider feeding tube placement.¹ Artificial nutrition in patients with advanced disease and dementia is rarely appropriate and often considered to be unacceptable by most patients.²

One of the most commonly used staging schemes for HD is the Shoulson and Fahn Total Functional Capacity Rating (TFC) Scale which identifies five stages of disease. It is estimated that HD patients experience an average decline of 0.7 points per year highlighting the slow progression of this disease. Late-stage HD (Stage 4 or 5) is considered 0-2 points, specifically when individuals require assistance in all activities of daily living and often are unable to remain at home.¹

Personal details in the case published have been altered to protect patient privacy.

For palliative care consultations please contact the Palliative Care Program at PUH/MUH, 647-7243, beeper 8511, Shadyside Dept. of Medical Ethics and Palliative Care, beeper 412-647-7243 pager # 8513, Perioperative/ Trauma Pain 647-7243, beeper 7246, UPCI Cancer Pain Service, beeper 644-1724, Interventional Pain 784-4000, Magee Women's Hospital, beeper 412-647-7243 pager #: 8510, VA Palliative Care Program, 688-6178, beeper 296. Hillman Outpatient: 412-692-4724. For ethics consultations at UPMC Presbyterian-Montefiore and Children's page 958-3844. With comments about "Case of the Month" call Dr. Robert Arnold at (412) 692-4834.



Discussion Continued

Total Functional Capacity Rating Scale ¹		
Domain	Ability	Score
Occupation	Unable	0
	Marginal work only	1
	Reduced capacity for usual job	2
	Normal	3
Finances	Unable	0
	Major assistance	1
	Slight assistance	2
	Normal	3
Domestic Chores	Unable	0
	Impaired	1
	Normal	2
Activities of Daily Living	Total care	0
	Gross tasks only	1
	Minimal impairment	2
	Normal	3
Care Level	Full-time nursing care	0
	Home for chronic care	1
	Home	2
TOTAL	Range 0-13	

TFC Total Score	Stage
11-13	I
7-10	II
3-6	III
1-2	IV
0	V

Late-stage HD can last for up to ten years, and so specific scales have been developed for advanced disease with TFC scale score of zero. An advanced HD TFC scale, modeled after the TFC scale, identifies five stages within advanced disease (Stage A1-A5). It does not account for mood, behavioral changes, or acute illness.¹

Advanced HD Functional Capacity Scale ¹					
	Mobility	Feeding	Contenance	Communication	Participate
4	Walks, may have missteps, but no more than occasional falls			Communicates with people other than family, caregivers	Able to participate actively in family/ residence activities
3	Frequent falls or very frequent near falls	Eats independently, using at least a fork or spoon	Continent of both stool and urine, takes care of toilet hygiene	Phrases or sentences only intelligible to family or in context	Able to participate in some or occasional activities
2	Wheelchair, independent	Uses a cup/straw, finger foods	Not always continent, or poor toilet hygiene	Single words or severe dysarthria; limited ability to speak even with family or in context	Able to attend some activities but little or no active participation
1	Able to sit but dependent on others for mobility	Must be fed	Incontinent most or all of the time but aware	Mute but attempts to communicate (grunts, screams)	Able to respond interactively in some way to others
0	Bed-bound or unable to sit	Most or all nutrition provided by feeding tube	Incontinent and unaware or passive about help	Mute	Non-interactive

Stage	Advanced TFC Total Score
A1	16-18 (largely independent)
A2	10-15 (still trying to be independent but not very successful)
A3	6-9 (receives assistance with all activities but at least interacts)
A4	2-5 (requires full assistance with everything)
A5	0-1 (terminal stages)

Personal details in the case published have been altered to protect patient privacy.

For palliative care consultations please contact the Palliative Care Program at PUH/MUH, 647-7243, beeper 8511, Shadyside Dept. of Medical Ethics and Palliative Care, beeper 412-647-7243 pager # 8513, Perioperative/ Trauma Pain 647-7243, beeper 7246, UPCI Cancer Pain Service, beeper 644 –1724, Interventional Pain 784-4000, Magee Women’s Hospital, beeper 412-647-7243 pager #: 8510, VA Palliative Care Program, 688-6178, beeper 296. Hillman Outpatient: 412-692-4724. For ethics consultations at UPMC Presbyterian-Montefiore and Children’s page 958-3844. With comments about “Case of the Month” call Dr. Robert Arnold at (412) 692-4834.



Discussion Continued

Late-stage symptom management is complex and generally involves a multidisciplinary approach including medications, occupational and physical therapy, nursing support, speech therapy and environmental changes. Environmental changes involve removing long cords and restraints that a patient could become tangled in, as well as other hazards that a patient may fall onto and injure themselves. Occupational therapists can help to optimize function, provide modified equipment for eating, identify heavy-duty furniture or appliances, and creatively pad the chair, bed, walls, and furniture corners. Physical therapists can help with early acclimation to wheelchairs or other assistive devices. Speech therapists can train patients and caregivers in safe feeding strategies, computer-based assistive communications, and word boards. Nursing support is critical for patient care and best provided by staff who are familiar with the patient, their likes and dislikes, and body language to anticipate needs or make an educated guess as to the HD person's concerns before the person fully loses the ability to interact.¹

Stage A5 using the Advanced TFC scale is often considered the terminal stage of disease. Morbidity and mortality are often related to oral-motor dysfunction and dysphagia. Symptoms and behaviors which may prompt enrollment in hospice care are non-directed screaming, weight loss with the inability or refusal to eat, mutism, limited or no ability to interact, and inability to ambulate.¹ In particular, non-directed screaming by a previously mute patient is often a sign that the patient has entered the terminal stage with a prognosis of a few months or less. This is generally interpreted as a patient experiencing physical or emotional discomfort and long acting opioids such as transdermal fentanyl or oral long-acting morphine are typically used and effective. Symptoms of agitation, spasticity or dystonia, and sialorrhea may worsen in the terminal phase and are treated with anxiolytics, muscle relaxants and anticholinergics respectively.¹

Back to the Case: Ms. P's Total Functional Capacity score was zero which translates to Stage 5. She scored two points, or Stage A4, on the advanced TFC scale. Her immobility, limited ability to speak or interact, and progressive weight loss made her a candidate for hospice enrollment. In discussions with the POA, it became clear that Ms. P would find her current quality of life unacceptable.

Ms. P had not wanted any life-prolonging measures, including a feeding tube, and her POA believed she would want to only focus on comfort. She volunteered her understanding that Ms. P's death may be the result of a secondary illness such as an aspiration pneumonia and preferred to treat her symptoms and avoid antibiotics which could potentially prolong her suffering. Palliative Care worked with the primary service to put a treatment plan in place that focused on her comfort and quality of life. Specifically, they discussed no longer using antibiotics for her recurrent aspiration pneumonia. A low-dose, long-acting opioid was started.

Ms. P was admitted to a skilled nursing facility with hospice care, requiring a single room with no bed or furniture, a mattress on the floor, and surrounding pads. Ms. P passed away approximately two months after admission to hospice care.

References:

1. Nance M, Paulsen JS, Rosenblatt A, Wheelock V. A Physician's Guide to the Management of Huntington's Disease. Third Edition. Huntington's Disease Society of America 2011.
2. Phillips W, Shannon KM, Barker RA. The Current Clinical Management of Huntington's Disease. *Mov Disord* 2008; 23:1491-1504.
3. Marks S, Hung S, Rosielle DA. Palliative Care for Patients with Huntington's Disease, Fast Fact 201. *J Palliat Med* 2016; 19: 394-399.

Personal details in the case published have been altered to protect patient privacy.

For palliative care consultations please contact the Palliative Care Program at PUH/MUH, 647-7243, beeper 8511, Shadyside Dept. of Medical Ethics and Palliative Care, beeper 412-647-7243 pager # 8513, Perioperative/ Trauma Pain 647-7243, beeper 7246, UPCI Cancer Pain Service, beeper 644-1724, Interventional Pain 784-4000, Magee Women's Hospital, beeper 412-647-7243 pager #: 8510, VA Palliative Care Program, 688-6178, beeper 296. Hillman Outpatient: 412-692-4724. For ethics consultations at UPMC Presbyterian-Montefiore and Children's page 958-3844. With comments about "Case of the Month" call Dr. Robert Arnold at (412) 692-4834.