

■ *The Movement Disorder*

INTRODUCTION

There are two parts to the movement disorder associated with Huntington disease: the presence of involuntary movements, and the impairment of voluntary movements. The involuntary movements are called chorea, or choreoathetosis, and consist of irregular jerking or writhing movements. Chorea is the most noticeable feature of HD. In fact, the condition is often referred to as Huntington's chorea, yet the impairment of voluntary movement is more highly correlated with functional disability. Abnormal eye movements (interrupted pursuit and slow, hypometric saccades), slow and uncoordinated fine movements, dysarthria, gait disturbance, and dysphagia can be largely independent of chorea and may limit a person's ability to work, care for himself, and communicate. Although it is tempting to treat the highly noticeable chorea of Huntington disease right away, it is important to remember that the drugs used to suppress chorea can have disadvantages of their own, including worsening of voluntary motor disturbance.

Table 2:

Principles Of Treatment Of The Movement Disorder

- Consider non-drug interventions first.
 - Pharmacologic treatment of chorea may worsen other aspects of the movement disorder, cognition, or mood.
 - Chorea may diminish over time, reducing the need for treatment.
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CHOREA

Many patients are not bothered by their chorea and may not even be aware of most of the movements. The physician and patient first need to establish whether the chorea requires any treatment at all. Is the chorea severe enough to interfere with voluntary activities such as writing, cooking, or eating? Does severe chorea seem to be causing falls or accidents? Is highly visible chorea a significant source of distress for the patient?

Before beginning medication for chorea, non-pharmacologic interventions should be considered. Chorea, like most forms of involuntary movement, is worsened by stress, anxiety, or depression, is decreased

during sleep, and often varies with posture or positioning. Treatment of underlying mood and anxiety disorders, and providing a calm, predictable environment are a first step. Various assistive devices may be helpful. These include padded, reclining chairs, padding for the bed, and wrist and ankle weights to reduce the amplitude of the chorea. Sources for some of these devices are provided in Appendix 3.

Doctor and patient also need to have realistic expectations for pharmacotherapy. Medications will not alter the progression of the underlying illness. They will not improve speech or the ability to swallow, prevent falls, or improve fine motor control. In fact, drug-related side effects such as sedation and rigidity may increase the risk of falls and decrease the intelligibility of speech. However, reduction of severe chorea may improve gross motor control and may be of cosmetic value.

Akathisia is an extremely uncomfortable internal sense of restlessness, sometimes induced by neuroleptics, which may cause patients to pace, or be unable to sit still. It can be mistaken for agitation or anxiety, prompting the physician to increase the dose of the offending drug, creating a vicious cycle.

The movement disorder of HD changes over time. In most patients chorea eventually peaks and then begins to decline, while rigidity and bradykinesia become more significant. At this point, the drugs that helped to suppress chorea may no longer be needed, and in fact may worsen HD-related rigidity. Therefore it is important to assess the need for anti-chorea medication at regular intervals, and perhaps to make periodic trials of dose reduction or discontinuation.

Table 3:

Medications Used To Suppress Chorea

Class	Medication	Starting Dose	Maximum Dose	Adverse Effects
Neuroleptics	Haloperidol	0.5–1mg/day	6–8mg/day	sedation, parkinsonism, dystonia, akathisia, hypotension, constipation, dry mouth, weight gain
	Fluphenazine	0.5–1mg/day	6–8mg/day	same
	Risperidone	0.5–1mg/day	6mg/day	less parkinsonism
	Thiothixene	1–2mg/day	10–20mg/day	less parkinsonism, more sedation and postural hypotension
	Thioridazine	10mg/day	100mg/day	similar to thiothixene
Benzodiazepines	Clonazepam	0.5mg/day	4mg/day	sedation, ataxia, apathy, withdrawal seizures
	Diazepam	1.25mg/day	20mg/day	same
Dopamine Depleting Agents	Reserpine	0.1mg/day	3mg/day	hypotension, sedation, depression
	Tetrabenazine	25mg/day	100mg/day	less hypotension

Three classes of medication are commonly used to suppress chorea in Huntington disease: neuroleptics, such as haloperidol and fluphenazine; benzodiazepines, such as clonazepam and diazepam; and dopamine depleting agents, such as reserpine and tetrabenazine. Each class has its advantages and disadvantages.

The suppression of movement, regarded as a side effect when neuroleptics are used to treat psychosis, is the desired effect when they are used to treat chorea. Therefore the most popular neuroleptic agents are the high potency drugs, which can also induce the most parkinsonism. Haloperidol and fluphenazine are most commonly prescribed. They should be started at a low dose, 0.5 to 1mg once or twice a day, and gradually increased to efficacy. Doses higher than 6–8mg per day have not generally been found helpful in treating chorea. Risperidone is a newer neuroleptic which does not cause as much parkinsonism as the other high potency agents, but is still useful in suppressing chorea and may relieve agitation as well. It may also be started at 0.5–1mg once or twice a day, with some patients tolerating doses as high as 6–8mg daily.

In some cases, patients who experience unacceptable rigidity, akathisia, or dystonia with high potency agents may benefit from a lower potency neuroleptic such as thiothixene or thioridazine. This may be preferable to adding an anticholinergic agent to the original drug to counteract the side effects. Lower potency agents tend to be more sedating, however, and are more inherently anticholinergic, producing more tachycardia, postural hypotension, constipation, and delirium. Thiothixene can be started at 1–2mg once or twice a day and increased to 10–20mg/day. Thioridazine, which is even lower potency, can be started at 10mg once or twice a day and increased to about 100mg/day.

Patients starting neuroleptics should be warned about two unlikely, but potentially serious adverse effects. The first is tardive dyskinesia, a syndrome of involuntary movements often first noted in the face and mouth, that develops in some patients taking neuroleptics. Tardive dyskinesia is of concern because the symptoms are usually permanent, and will likely be hard to recognize in someone with HD. The other serious problem is neuroleptic malignant syndrome, a rare, but life threatening reaction characterized by acute onset of delirium, rigidity, and fever, often accompanied by leukocytosis, and elevated CPK. Families should know about this so that the patient can be given prompt medical attention if it develops.

Benzodiazepines, such as clonazepam and diazepam can also be useful in the treatment of chorea. Some clinicians prefer them to neuroleptics because they do not induce parkinsonism or tardive dyskinesia. Sedation and the increased risk of delirium are the main deleterious side effects, along with tolerance, withdrawal symptoms, and the potential for abuse. Long acting varieties such as clonazepam and diazepam are favoured because they require less frequent dosing, provide more even coverage of symptoms throughout the day, and are less likely to precipitate withdrawal symptoms if a dose is missed. Clonazepam may be started at 0.5mg per day, and may be raised as high as 4mg per day, in divided doses. Diazepam may be dosed from about 1.25mg to 20mg per day, also in divided doses.

Some clinicians favour dopamine depleting agents as a treatment for chorea. While these drugs do share some of the “neuroleptic” side effects, they may be milder at low doses, and they have not been shown to cause tardive dyskinesia. The class includes reserpine and tetrabenazine. Reserpine was used in the past as an antihypertensive, and may cause hypotension. This can be minimized by giving the drug at bedtime.

Parkinsonism, restlessness, dizziness, and sedation are other common side effects. The increased rate of depression in patients taking these agents is also of concern. Reserpine may be started at 0.1mg per day and increased weekly to a dose as great as 3mg per day. Tetrabenazine is similar in action to reserpine, but is felt by some clinicians to be more effective and is less likely to cause hypotension. It can be started at 12.5mg bid or tid and increased over several weeks to a maximum of 75 or 100mg per day in divided doses.

RIGIDITY, SPASTICITY, AND DYSTONIA

Rigidity and spasticity tend to emerge later in the course of Huntington disease, except in cases of childhood onset, in which they are often present from the beginning. They can impair gait, lead to falls, and necessitate the use of a wheelchair. Dystonia may include twisting, tilting or turning of the neck (torticollis), involuntary arching of the back (opisthotonos) and arching of the feet. It may be a symptom of HD, or a side effect of neuroleptic therapy.

A variety of medications have been used to treat rigidity, spasticity, and dystonia, all with modest success at best. Benzodiazepines, such as clonazepam, or baclofen, starting at 10mg/day and increasing up to 60mg may relieve stiffness, but may also increase bradykinesia. Tizanidine, a clonidine like drug, is sometimes helpful for spasticity, beginning with 2mg qhs and increasing every 4–7 days to a maximum of 12–24mg in divided doses. Antiparkinsonian medicines such as amantadine 50–200mg/day, levodopa/carbidopa 25/100mg two to three times per day, or bromocriptine beginning at 1.25mg bid, increasing every few weeks, may be helpful with bradykinesia or rigidity, and some clinicians have tried trihexyphenidyl, 2–5mg, bid to tid. All of these medicines may cause delirium and may lose their efficacy after several months. Consultation with a physiotherapist or physiatrist to design a program to mobilize the patient and prevent contractures may be an important component to the management of rigidity and spasticity. Botulinum toxin injections have been used rarely, but might be beneficial if severe rigidity of a small muscle or group of muscles is disturbing function.

MYOCLONUS, TICS, AND EPILEPSY

Myoclonus, sudden brief jerks involving groups of muscles, is more common in juvenile-onset HD, where it may be mistaken for a seizure. Like chorea, myoclonus may not be disabling or particularly distressing, but may respond to treatment with clonazepam or divalproex sodium if treatment is necessary. Tics are brief, intermittent stereotyped movements such as blinking, nose twitching, head jerking, or transient abnormal postures. Tics which involve the respiratory and vocal apparatus may result in sounds including sniffs, snorts, grunts, coughs, and sucking sounds. Patients may be unaware of vocal tics, but family members may find the incessant noises grating. They should be helped to understand that the tics are not under voluntary

control. Tics generally do not by themselves require treatment, but may respond to neuroleptics, benzodiazepines, or SSRIs.

Epilepsy is uncommon, though not unheard of, in adults with HD, but is said to be present in 30% of individuals with juvenile-onset HD. A first seizure in an HD patient should not be attributed to HD without further evaluation as it may be indicative of an additional neurologic problem, such as a subdural hematoma sustained in a fall. The workup of a first seizure should include a complete exam, laboratory studies to rule out an infection or metabolic disturbance, an EEG, and a brain imaging study. The treatment of a seizure disorder in a person with HD depends on the nature of the seizures. In the juvenile HD patient, myoclonic epilepsy or other generalized seizures may suggest divalproex sodium as a first treatment choice. Although seizure management in HD is not usually difficult, for the occasional patient seizure control is quite difficult to achieve, requiring multiple medications or specialized referral.

SWALLOWING DIFFICULTIES

Dysphagia is, directly or indirectly, the most common cause of death in people with late stage HD, whether through choking, aspiration, or malnutrition. Dysphagia results from impaired voluntary control of the mouth and tongue, impaired respiratory control due to chorea, and impaired judgement, resulting in eating too rapidly, or taking overly large bites of food and gulps of liquid. Dry mouth, which can be brought on by neuroleptics, antidepressants, and anticholinergics, may worsen the problem.

No medications are known to improve swallowing directly. Early referral to a speech-language pathologist will help to identify swallowing difficulties, and periodic reassessment can identify changes in swallowing ability and suggest appropriate non-pharmacologic interventions, such as a change in food consistency. Devices such as enlarged grips for silverware and nonslip plates with raised edges to prevent spilling may prolong independent eating. HD affected individuals should be instructed early in the disease, before the onset of dysphagia, to eat slowly and deliberately, to sit in an upright position during and after meals, to take small bites, and to clear the mouth of food after each bite by taking sips of liquid.

Individuals with dysphagia should avoid doing other activities while eating, in order to concentrate on chewing and swallowing. For instance, patients should not talk while eating, nor be distracted by television or ambient noise. Those who tend to hyperextend the neck due to chorea or dystonia should be encouraged and reminded to use a "chin-tuck" position. Drinking fluid through a straw may be easier than drinking directly from a cup, and the use of a covered cup or mug, like a "sippy cup" used by young children, may prevent spillage due to chorea. Grainy items, such as ground beef or rice, may irritate the pharynx and cause choking. Foods such as steak, which are hard to chew, should also be avoided, or ground to a puree. Patients may have difficulty adjusting to different textures of food, and may do better if they finish each item on the plate in turn.

Table 4:

Swallowing Tips

- Eat slowly and without distractions.
 - Prepare foods with appropriate size and texture.
 - Eating may need to be supervised.
 - Caregivers should know the Heimlich manoeuvre.
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In late HD, when even liquids may be difficult to swallow, the texture of food should be soft and smooth, and liquids may be thickened with an additive (see Appendix 3). For those patients who may be unable to follow instructions reliably, a caregiver can cut the food in advance, and ensure that each mouthful has been completely chewed and swallowed before the next bite is begun. Supervision throughout the meal may be necessary, and the family or caregiver should be taught to perform the Heimlich manoeuvre.

In some cases, eating eventually requires so much energy and concentration that the patient becomes tired and frustrated before consuming adequate amounts of food. Weight loss, very prolonged mealtimes or an inability to handle utensils may be the signal that he will need to be fed for at least part of the meal. Self-feeding may be prolonged by having the patient eat more frequent, but smaller meals, and by using "finger foods." The transition to assisted feeding does not have to be all or nothing, as patients may still be able to eat unassisted at certain times and be fed at other times.

Choking may decrease once self-feeding is stopped, because the caregiver will have greater control over the size and frequency of the bites. The caregiver should still promote eating slowly, and not talking while eating, and should make sure the mouth is empty before each bite. With supervision, most patients are able to assist with feeding and to take adequate amounts of food by mouth quite far into the illness. However, before dysphagia and communication difficulties become severe, the issue of feeding tubes should be discussed with the patient and family, to ensure that appropriate nutrition can be maintained throughout the illness. A gastrostomy tube can clearly improve nutritional status in a debilitated person with severe dysphagia, and may prolong life. However, patients and families may not desire this intervention late in the course of HD. The question of whether to use a gastrostomy tube, and other end of life issues are discussed in the final section of chapter 6.

NUTRITION

Weight loss is a common problem in Huntington disease. This is probably due in part to diminished food intake because of dysphagia, fatigue, and depression. However many HD patients also require a large caloric intake to maintain their body weight. This may be simply due to the expenditure of energy through involuntary movements, but there may be other metabolic reasons not fully understood. Two strategies can be employed to increase the caloric intake of someone with HD: increase the number of meals, or increase the calorie content of the food. The first goal can be achieved by eating five small meals a day or by adding high calorie snacks such as milkshakes. The caloric content of the food can be increased by measures such as adding oil to soups, drinking cream instead of skim milk, adding margarine liberally as a condiment, and focusing on easily eaten, high-calorie foods such as pasta with cream based sauce. Consultation with a nutritionist can help in selecting the most appropriate foods and supplements to meet the patient's needs. Regaining lost weight sometimes results in improved alertness and responsiveness, and often appears to reduce chorea as well. Maintaining hydration is also very important, particularly in the summertime in patients who may not be able to request fluids. Cyproheptadine, an antihistamine, given as 4mg at bed-time, may help increase weight by stimulating appetite in some patients.

DYSARTHRIA

Dysarthria, a difficulty with the physical production of speech, results largely from impairment of voluntary movement. Speech becomes slurred, dysrhythmic, variable in volume due to inconsistent breath support, and increasingly difficult to understand. Furthermore, just as patients do not always appreciate the presence or degree of chorea, some patients do not seem to be aware of distortions in their speech. For others, articulation is a constant source of frustration. No medications are known to be helpful, and dysarthria may be worsened by agents which suppress chorea. However, several interventions may enhance communication in these patients. The listener must do everything possible to promote successful communication, beginning with allowing enough time. Many HD patients thought to be incapable of communication can be understood if the listener is patient enough. Patients may need to be moved to a quieter, calmer environment, and urged to speak slowly. Patients can be asked to spell difficult to understand words. A communication board can also be useful in some cases. A speech-language pathologist may be able to provide additional insights and management strategies.

Dysarthria may be compounded by cognitive problems found in HD, such as word-finding difficulty, difficulty initiating speech, or difficulty completing a sentence. Even those with severe cognitive impairments often respond to cues, such as asking for the size, shape or color of an object. Even severely impaired patients may be able to respond accurately to a series of yes and no questions. If unsuccessful attempts at communication become very frustrating, it may be better to take a break. The desire for social interaction generally remains, even in those with advanced HD, so strategies for communication should be a priority.

Table 5:

Coping Strategies For Communication

- Allow the person enough time to answer questions.
 - Offer cues and prompts to get the person started.
 - Give choices. For example, rather than asking “what do you want for dinner?” ask “do you want hamburgers or meatloaf?”
 - Break the task or instructions down into small steps.
 - If the person is confused, speak more simply and use visual cues to demonstrate what you are saying.
 - Ask the person to repeat phrases you did not understand, or spell the words.
 - Alphabet boards, yes-no cards, or other communication devices may be helpful.
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FALLS

Falls are common in persons with HD, and can be a source of significant morbidity. Usually seen more in the moderate to advanced stages, they often result from the combination of spasticity, rigidity, chorea, and loss of balance. Pharmacotherapy to prevent falls could include treatment of chorea, rigidity, spasticity and dystonia, while minimizing the use of drugs such as neuroleptics and benzodiazepines, whose side effects include sedation, ataxia, or parkinsonism. Most efforts at prevention, however, involve not drugs, but modification of the environment and behaviour of the patient. Occupational therapists and physiotherapists can instruct patients in how to sit, stand, transfer, and walk more safely. Installing handrails in key loca-

tions, and minimizing the use of stairs can help to reduce falls. Some families convert a ground floor office or den into a bedroom. Furniture such as tables and desks, particularly items with sharp corners, should be arrayed along the periphery of the room, where they will present less of an obstacle. Floors should be carpeted to lessen the impact when falls do occur. Patients who fall out of bed may have a mattress placed beside the bed at night, or may sleep on a mattress placed directly on the floor.

HD patients will eventually become unable to walk and will need to be transported in a wheelchair. A weighted and padded chair, perhaps with a wedge to keep the hips tilted, or a pommel between the legs, may minimize the chance of a severely choreic or dystonic patient falling or sliding out, or knocking over the chair (see Appendix 3). Use of a wheelchair is not an all or nothing proposition. Mobility may be extended by using the wheelchair for longer excursions and using other assistive devices such as a walker for shorter distances, or in the home. Walkers with front wheels may be particularly useful when rigidity or loss of balance is a problem. Patients who are particularly prone to falls sometimes wear helmets, or elbow and knee pads to minimize injury. Physiotherapy may also help by teaching patients how to minimize injury in a fall and how to get up again after a fall.

GENERAL SAFETY MEASURES

A number of other environmental interventions may reduce the risk of injury. Patients who smoke should do so in a room without flammables, such as rugs, curtains and overstuffed furniture. Patients may need to stop using sharp knives and to switch to microwave cooking to prevent burns and spills. Falls in the bathroom are particularly dangerous, but there are a variety of assistive devices that can be installed. Consultation with a visiting nurse, or a visit from a physiotherapist or occupational therapist may be very helpful for any mid-stage HD patient being cared for in the home. A sample home visit consultation form is provided in Appendix 4.