Huntington Chorea

Last Updated: March 9, 2005

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Synonyms and related keywords: Huntington disease, Huntington's disease, HD, Huntington's chorea, HC, chorea, choreiform movements, progressive dementia, involuntary movements, rigidity, huntington, progressive neurological disorder, irritability, moodiness, antisocial behavior, piano-playing motion of fingers, facial grimaces, dancing gait, choreoathetoid movements, weight loss, behavioral problems, depression, dementia, seizures, movement disorder, facial twitching, twitching and writhing of distal extremities, fast eye movements, saccades, dystonia, impaired cognitive abilities

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Disclosure

INTRODUCTION

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Background: Huntington disease (HD), also known as Huntington chorea (HC), is an inherited disease characterized by choreiform movements and progressive dementia.

In adults, HD most often causes involuntary movements, but rigidity can also be a feature of the disease.

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Patient Education

The initial diagnosis is rarely established in the emergency department, but patients with established disease may present to the ED because of worsening symptoms.

Pathophysiology: HD is inherited as an autosomal dominant disorder with complete penetrance. An HD gene has been identified with an abnormal protein product (huntingtin) that can be identified in the brain. The link between this protein and the selective loss of neuronal groups in the CNS remains to be established. HD has now been identified genetically as a trinucleotide CAG-repeat mutation on chromosome 4. The CAG repeat length may be important in determining the age of onset and the rate of disease progression.

Frequency:

- **In the US:** Prevalence of HD in the United States is 5.15 cases per 100,000 persons.
- **Internationally:** HD is encountered throughout the world; however, localized geographic clusters of disease exist. Countries that have been settled by western Europeans have an incidence of the disease similar to the incidence in the United States.

Mortality/Morbidity: HD is a progressive neurological disorder usually leading to death 15-20 years after onset of neurological or psychological impairment.

Race: HD is found in all ethnic groups.

Sex: Males and females are diagnosed in equal numbers.

Age:

- Symptoms arising from a typical presentation of HD usually do not develop until a person is aged 35 years or older. By the time of diagnosis, many patients already have had children and have passed the gene to another generation.
- As many as 10% of patients with HD have a juvenile form of the disease in which the onset of symptoms may occur when the patient is younger than 20 years.
- Muscular rigidity is more common with juvenile-onset illness.

Dementia Center

Huntington Disease Dementia Overview

Huntington Disease Dementia Causes

Huntington Disease Dementia Symptoms

Huntington Disease Dementia Treatment

History:

- Between one half and three fourths of patients affected by HD present with primary complaints of involuntary movements or rigidity. In the remainder of cases, the primary presentation is one of early mental status changes that initially appear as increased irritability, moodiness, or antisocial behavior.
- Classic choreiform movements begin as a piano-playing motion of the fingers or as facial grimaces.
- As the disorder advances and involves the trunk, a characteristic dancing gait evolves. Although patients appear to be off-balance, the ability to balance is actually well preserved.
- Patients who develop HD by the time they are aged 35 years often become bedridden within 15-20 years.
- A dramatic weight loss may occur due to energy expenditure from choreoathetoid movements.
- Behavioral problems may be the first noticeable issues. Depression often occurs early in the course of the illness.
- Patients may be argumentative, impulsive, or erratic.
- Dementia develops as the disease progresses.

- Childhood cases tend to present with rigidity rather than with chorea.
- Younger patients may develop seizures.
- Family history is of paramount importance in making the diagnosis.
    - If a patient with movement disorder or dementia is known to have a parent with HD, the diagnosis of HD is highly likely.
    - The typical clinical picture of HD may appear in patients who have no family history of the disease. Whether the disease actually can arise de novo through new genetic mutations is unclear.

Physical:

- The pathognomonic feature of HD is the movement disorder.
    - Chorea is the most common movement disorder. In HD, chorea often appears as facial twitching or as twitching and writhing of the distal extremities.
    - Fast eye movements (ie, saccades) often are impaired.
    - As HD disease progresses, the movement disorder becomes more generalized. Eventually, the patient's gait is impaired.
    - Rigidity and dystonia predominate in later stages of the disease in adults. In juvenile cases, rigidity and dystonia may appear as the initial symptoms.
    - Symptoms become worse with anxiety or stress.
- A mental status examination may reveal depression.
- Impaired cognitive abilities may be detected on physical examination.

Causes:

- HD is a genetic disease inherited as an autosomal dominant trait. The gene is present on chromosome 4.
    - The gene encodes a protein known as huntingtin. Gradually, this protein accumulates within brain cells.
    - Brain cells are damaged when levels of huntingtin rise.
- Common medical opinion holds that nearly all cases are inherited and that appearance of the disease through new mutations is extremely rare.
- When a patient presents with HD, identification of a parent with an intermediate allele who lacks the full clinical manifestations of the disease is often possible.
- Family history is the principal identified risk factor.
- A small number of patients exist who have DNA-proven HD but a completely negative family history.

Other Problems to be Considered:

Hereditary nonprogressive chorea

Neuroacanthocytosis
Wilson disease
Ataxia-telangiectasia
Lesch-Nyhan syndrome
Hallervorden-Spatz disease
Fahr disease
Sydenham chorea
Drug-induced movement disorder
Chorea gravidarum
Hyperglycemic nonketotic encephalopathy
Hemichorea/hemiballism with subthalamic nucleus lesion
Periarteritis nodosa
Senile chorea
Essential chorea
Parkinson disease
Alzheimer disease
Creutzfeldt-Jakob disease
Pick disease
Bipolar disorder
Alcoholism
Antisocial personality disorder

WORKUP

Lab Studies:

- Some laboratories now offer DNA analysis to identify the *HD* gene. The DNA test is highly reliable and can confirm or refute the diagnosis in most cases.
- Because HD often has such a devastating course, DNA testing of family members who lack symptoms (predictive testing) is controversial. Careful counseling and follow-up care are essential if asymptomatic family members are to be tested. Interpretation may be problematic. Referral to a regional HD center or to an experienced neurologist for pretest and posttest counseling is recommended.
- Decreased endogenous gamma-aminobutyric acid (GABA), decreased choline-acetyltransferase, and decreased glutamic acid decarboxylase may be detected.

Imaging Studies:

- CT scan or MRI may show loss of a normally convex bulge of the caudate nucleus into the lateral ventricles. In patients with typical symptoms, this CT scan finding strongly supports the diagnosis of HD.
- Enlarged lateral ventricles usually are visible on head CT scan.

Other Tests:

- Familial screening for the disease is possible, but widespread testing awaits resolution of social and ethical considerations.

TREATMENT

Emergency Department Care:

- Dopamine receptor-blocking agents (eg, phenothiazines, haloperidol) may be helpful in reducing some of the involuntary movements, but such therapy always should be coordinated with the primary care provider.

Consultations: Consultation with a neurologist is appropriate if the diagnosis is in question. Behavioral problems may require consultation with a primary care giver or a psychiatrist.

- Affective disorders, anxiety disorders, and psychosis may occur in patients with Huntington disease. The initial pharmacologic approach to

treatment is similar to that of other patients.

- Drug-resistant depression may yield to electroconvulsive therapy.

No specific pharmacologic therapy for HD exists. Chorea sometimes may be suppressed with drugs, but the associated adverse effects can be severe. Most patients are actually untroubled by the choreic movements. Suppression of the movements does not result in improved function.

Depression is common and may require treatment. Serotonin reuptake inhibitors or cyclic antidepressants may be tried. Irritability and explosive behavior are problems in some patients. Carbamazepine or valproate may be useful, but the patient's primary physician or neurologist should coordinate such therapy.

Drug Category: *Major tranquilizer* -- May suppress chorea but will be accompanied by sedation, anticholinergic adverse effects, or rigidity.

Drug Name	Haloperidol (Haldol) -- Useful in the treatment of irregular spasmodic movements of the limbs or facial muscles.
Adult Dose	2.5-10 mg/d PO divided bid/tid
Pediatric Dose	0.5 mg/kg/d PO divided bid/tid
Contraindications	Documented hypersensitivity; narrow-angle glaucoma; bone marrow suppression; severe cardiac or liver disease; severe hypotension; subcortical brain damage
Interactions	May increase tricyclic antidepressant serum concentrations and hypotensive action of antihypertensive agents; phenobarbital or carbamazepine may decrease effects of haloperidol; coadministration with anticholinergics may increase intraocular pressure; encephalopathy-like syndrome associated with concurrent administration of lithium and haloperidol
Pregnancy	D - Unsafe in pregnancy

Precautions	Severe neurotoxicity manifesting as rigidity, or inability to walk or talk may occur in patients with thyrotoxicosis also receiving antipsychotics; if given IV/IM, watch for hypotension; caution in diagnosed CNS depression or cardiac disease; if history of seizures, benefits must outweigh risks; significant increase in body temperature may indicate intolerance to antipsychotics (discontinue if occurs)

Further Inpatient Care:

- Admission for supportive care occasionally is necessary.
- If depression is disabling, a psychiatric admission may be appropriate.

Further Outpatient Care:

- Refer patient to the primary care physician or neurologist if symptoms progress.

Transfer:

- Transfer is seldom necessary.

Deterrence/Prevention:

- The issue of family testing for the *HD* gene is controversial. Some may use the information to alter reproductive choices.
- Referral for genetic counseling may be considered for families.

Complications:

- The dementia eventually progresses in every patient and may be disabling.
- Patients may develop difficulty swallowing and may suffer choking episodes.
- Chorea or rigidity nearly always is present.
- Personality changes may be prominent in some patients.
- Patients with depression may be at risk for suicide.

Prognosis:

- The disease is slowly but inexorably progressive.
- Typical life expectancy is 10-25 years after diagnosis.

Patient Education:

- The Huntington Disease Society of America (HDSA) is a source of information for the public (1-800-345-HDSA). Many local chapters exist.
- Genetic counseling may be considered for family members, especially those who wish to know their chances of inheriting HD. While true that the disease develops if the gene is inherited, the odds of inheritance are 50-50.
- EDs are poor venues for counseling. Defer counseling to a setting that allows confirmation of the diagnosis, follow-up care, and genetic counseling.
- For excellent patient education resources, visit eMedicine's Dementia Center. Also, see eMedicine's patient education article Huntington Disease Dementia.

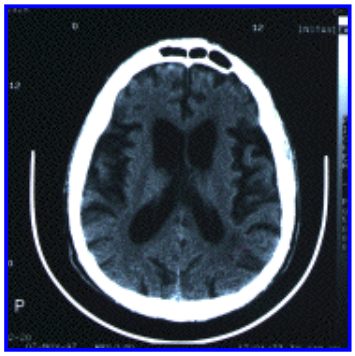
MISCELLANEOUS

Medical/Legal Pitfalls:

- A failure to rule out reversible forms of dementia and reversible movement disorders when considering a diagnosis of Huntington disease invites problems.
- No rapid ED test for the definitive diagnosis of HD exists. A failure to refer a patient who has an initial HD presentation to a neurologist invites problems.

PICTURES

Caption: Picture 1. The CT scan is of a patient with advanced Huntington disease. Note the diffuse cortical atrophy and the selective atrophy of the caudate nuclei as shown by the excessive width of the lateral ventricles.



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Picture Type: CT

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Huntington Chorea excerpt