Huntington's disease

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Symptoms & causes Diagnosis & treatment Doctors & departments Care at Mayo Clinic

Diagnosis

Print

A preliminary diagnosis of Huntington's disease is based primarily on your answers to questions, a general physical exam, a review of your family medical history, and neurological and psychiatric examinations.

Neurological examination

The neurologist will ask you questions and conduct relatively simple tests in the office to judge:

Motor symptoms

- Reflexes
- Muscle strength
- Muscle tone
- Coordination
- Balance

Sensory symptoms

- Sense of touch
- Vision and eye movement

- Hearing

Psychiatric symptoms

- Mental status
- Mood

Neuropsychological testing

The neurologist may also perform standardized tests to assess:

- Memory
- Reasoning
- Mental agility
- Language function
- Spatial reasoning

Psychiatric evaluation

You'll likely be referred to a psychiatrist for an examination to judge a number of factors that could contribute to your diagnosis, including:

- Emotional state
- Patterns of behaviors
- Quality of judgment
- Coping skills
- Signs of disordered thinking
- Evidence of substance abuse

Brain imaging and function

Your doctor may order brain-imaging tests for assessing the structure or function of the brain. The imaging technologies may include magnetic resonance imaging (MRI) or computerised tomography (CT) scans that provide detailed images of brain structures.

These images may reveal structural changes at particular sites in the brain affected by Huntington's disease, although these changes may not be apparent early in the course of the disease. These tests can also be used to rule out other conditions that may be causing symptoms.

Genetic counseling and testing

If symptoms strongly suggest a diagnosis of Huntington's disease, your doctor may recommend a genetic test for the defective gene.

This test can confirm the diagnosis, and it may be valuable if there's no known family history of Huntington's disease or if no other family member's diagnosis was confirmed with a genetic test. But the test won't provide information that might help determinine a treatment plan.

Before undergoing such a test, the genetic counselor will explain the benefits and drawbacks of learning test results. The genetic counselor can also answer questions about the inheritance patterns of Huntington's disease.

Predictive genetic test

A genetic test can be given to someone who has a family history of the disease but shows no signs or symptoms. This is called predictive testing. The test result has no treatment benefit, and it doesn't indicate when disease onset will begin or what symptoms are likely to appear first.

Some people may elect to do the test because they find it more stressful not knowing. Others may want to take the test before they make decisions about having children.

Risks may include problems with insurability or future employment and the stresses of facing a fatal disease. In principle, federal laws exist that make it illegal to use genetic testing information to discriminate against people with genetic diseases.

These tests are only performed after consultation with a genetic counselor.

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EEG (electroencephalogram)

Genetic testing

Treatment

No treatments can alter the course of Huntington's disease. But medications can lessen some symptoms of movement and psychiatric disorders. And multiple interventions can help a person adapt to changes in his or her abilities for a certain amount of time.

Medication management is likely to evolve over the course of the disease, depending on the overall treatment goals. Also, drugs to treat some symptoms may result in side effects that worsen other symptoms. Therefore, the treatment goals and plan will be regularly reviewed and updated.

Medications for movement disorders

Drugs to treat movement disorders include the following:

- **Tetrabenazine** (Xenazine) is specifically approved by the Food and Drug Administration to suppress the involuntary jerking and writhing movements (chorea) associated with Huntington's disease. A serious side effect is the risk of worsening or triggering depression or other psychiatric conditions.

  Other possible side effects include drowsiness, nausea and restlessness.

- **Antipsychotic drugs,** such as haloperidol (Haldol) and chlorpromazine, have a side effect of suppressing movements. Therefore, they may be beneficial in treating chorea. However, these drugs may worsen involuntary contractions (dystonia) and muscle rigidity.

  Other drugs, such as risperidone (Risperdal) and quetiapine (Seroquel), may have fewer side effects but still should be used with caution, as they may also worsen symptoms.

- **Other medications** that may help suppress chorea include amantadine, levetiracetam (Keppra, others) and clonazepam (Klonopin). At high doses, amantadine can worsen the cognitive effects of Huntington's disease. It may also cause leg swelling and skin discoloration.

  Side effects of levetiracetam include nausea, stomach upset and mood swings. Clonazepam may worsen the cognitive side effects of Huntington's disease and cause drowsiness. It also has a high risk of dependence and abuse.

Medications for psychiatric disorders

Medications to treat psychiatric disorders will vary depending on the disorders and symptoms. Possible treatments include the following:

- **Antidepressants** include such drugs as citalopram (Celexa), escitalopram (Lexapro), fluoxetine (Prozac, Sarafem) and sertraline (Zoloft). These drugs may also have some effect on treating obsessive-compulsive disorder. Side effects may include nausea, diarrhea, drowsiness and low blood pressure.
- **Antipsychotic drugs** — such as quetiapine (Seroquel), risperidone (Risperdal) and olanzapine (Zyprexa) — may suppress violent outbursts, agitation, and other symptoms of mood disorders or psychosis. However, these drugs may cause different movement disorders themselves.
- **Mood-stabilizing drugs** that can help prevent the highs and lows associated with bipolar disorder include anticonvulsants, such as valproate (Depacon), carbamazepine (Carbatrol, Epitol, Tegretol) and lamotrigine (Lamictal).

Psychotherapy

A psychotherapist — a psychiatrist, psychologist or clinical social worker — can provide talk therapy to help a person manage behavioral problems, develop coping strategies, manage expectations during progression of the disease and facilitate effective communication among family members.

Speech therapy

Huntington's disease can significantly impair control of muscles of the mouth and throat that are essential for speech, eating and swallowing. A speech therapist can help improve your ability to speak clearly or teach you to use communication devices — such as a board covered with pictures of everyday items and activities. Speech therapists can also address difficulties with muscles used in eating and swallowing.

Physical therapy

A physical therapist can teach you appropriate and safe exercises that enhance strength, flexibility, balance and coordination. These exercises can help maintain mobility as long as possible and may reduce the risk of falls.

Instruction on appropriate posture and the use of supports to improve posture may help lessen the severity of some movement problems.

When the use of a walker or wheelchair is required, the physical therapist can provide instruction on appropriate use of the device and posture. Also, exercise regimens can be adapted to suit the new level of mobility.

Occupational therapy

An occupational therapist can assist the person with Huntington's disease, family members and caregivers on the use of assistive devices that improve functional abilities. These strategies may include:

- Handrails at home
- Assistive devices for activities such as bathing and dressing
- Eating and drinking utensils adapted for people with limited fine motor skills

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Psychotherapy

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Lifestyle and home remedies

Managing Huntington's disease is demanding on the person with the disorder, family members and other in-home caregivers. As the disease progresses, the person will become more dependent on caregivers. A number of issues will need to be addressed, and strategies to cope with them will evolve.

Eating and nutrition

Factors regarding eating and nutrition include the following:

- **People with Huntington's disease often have difficulty maintaining a healthy body weight.** Difficulty eating, higher caloric needs due to physical exertion or unknown metabolic problems may be the cause. To get adequate nutrition, more than three meals a day or the use of dietary supplements may be necessary.
- **Difficulty with chewing, swallowing and fine motor skills can limit the amount of food you eat and increase the risk of choking.** Problems may be minimized by removing distractions during a meal and selecting foods that are easier to eat. Utensils designed for people with limited fine motor skills and covered cups with straws or drinking spouts also can help.

Eventually, a person with Huntington's disease will need assistance with eating and drinking.

Managing cognitive and psychiatric disorders

Family and caregivers can help create an environment that may help a person with Huntington's disease avoid stressors and manage cognitive and behavioral challenges. These strategies include:

- Using calendars and schedules to help keep a regular routine
- Initiating tasks with reminders or assistance
- Prioritizing or organizing work or activities
- Breaking down tasks into manageable steps
- Creating an environment that is as calm, simple and structured as possible
- Identifying and avoiding stressors that can trigger outbursts, irritability, depression or other problems
- For school-age children or adolescents, consulting with school staff to develop an appropriate individual education plan
- Providing opportunities for the person to maintain social interactions and friendships as much as possible

Coping and support

A number of strategies may help people with Huntington's disease and their families cope with the challenges of the disease.

Support services

Support services for people with Huntington's disease and families include the following:

- **Nonprofit agencies,** such as the Huntington's Disease Society of America, provide caregiver education, referrals to outside services, and support groups for people with the disease and caregivers.
- **Local and state health or social service agencies** may provide daytime care for people with the disease, meal assistance programs or respite for caregivers.

Planning for residential and end-of-life care

Because Huntington's disease causes the progressive loss of function and death, it's important to anticipate care that will be needed in the advanced stages of the disease and near the end of life. Early discussions about this type of care enable the person with Huntington's disease to be engaged in these decisions and to communicate his or her preferences for care.

Creating legal documents that define end-of-life care can be beneficial to everyone. They empower the person with the disease, and they may help family members avoid conflict late in the disease progression. Your doctor can offer advice on the benefits and drawbacks of care options at a time when all choices can be carefully considered.

Matters that may need to be addressed include:

- **Care facilities.** Care in the advanced stages of the disease will likely require in-home nursing care or care in an assisted living facility or nursing home.
- **Hospice care.** Hospice services provide care at the end of life that helps a person approach death with as little discomfort as possible. This care also provides support and education to the family to help them understand the process of dying.
- **Living wills.** Living wills are legal documents that enable a person to spell out care preferences when he or she isn't able to make decisions. For example, these directions might indicate whether or not the person wants life-sustaining interventions or aggressive treatment of an infection.
- **Advance directives.** These legal documents enable you to identify one or more people to make decisions on your behalf. You may create an advance directive for medical decisions or financial matters.

Preparing for your appointment

If you have any signs or symptoms associated with Huntington's disease, you'll likely be referred to a neurologist after an initial visit to your family doctor.

A review of your symptoms, mental state, medical history and family medical history can all be important in the clinical assessment of a potential neurological disorder.

What you can do

Before your appointment, make a list that includes the following:

- Signs or symptoms — or any changes from "normal" — that may be causing concern
- Recent changes or stresses in your life
- All medications — including over-the-counter drugs and dietary supplements — and doses you take
- Family history of Huntington's disease or other disorders that may cause movement disorders or psychiatric conditions

You may want a family member or friend to accompany you to your appointment. This person can provide support and offer a different perspective on the effect of symptoms on your functional abilities.

What to expect from your doctor

Your doctor is likely to ask you a number of questions, including the following:

- When did you begin experiencing symptoms?
- Have your symptoms been continuous or intermittent?
- Has anyone in your family ever been diagnosed with Huntington's disease?
- Has anyone in your family been diagnosed with another movement disorder or psychiatric disorder?
- Are you having trouble performing work, schoolwork or daily tasks?
- Has anyone in your family died young?
- Is anyone in your family in a nursing home?
- Is anyone in your family fidgety or moving all the time?
- Have you noticed a change in your general mood?
- Do you feel sad all of the time?
- Have you ever thought about suicide?

By Mayo Clinic Staff

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