Huntington's Disease

Huntington's disease (HD) results from genetically programmed degeneration of brain cells (*neurons*) in certain areas of the brain. This degeneration causes uncontrolled movements, loss of intellectual faculties, and emotional disturbance.

CAUSES

HD is a familial disease. It is passed from parent to child through a mutation in the normal gene. Each child of an HD parent has a 50-50 chance of inheriting the HD gene. If a child does not inherit the HD gene, he or she will not develop the disease. Also, he/she cannot pass it to future generations. But a person who inherits the HD gene will sooner or later develop the disease. Whether one child inherits the gene has no bearing on whether others will inherit it

SYMPTOMS

Some early problems (*symptoms*) include:

- Mood swings.
- Depression.
- Irritability.
- Trouble with the following:
  - Driving.
  - Learning new things.
  - Remembering a fact.
  - Making decisions.

As the disease progresses, concentration on intellectual tasks becomes difficult. The patient may have difficulty feeding himself or herself and swallowing. The rate of disease progression and the age of onset vary from person to person.

DIAGNOSIS

Caregivers may diagnose HD by using:

- A genetic test.
- A complete medical history.
- Neurological and laboratory tests.

Presymptomatic testing is available for those who are at risk for carrying the HD gene. In 1 to 3 percent of individuals with HD, no family history of HD can be found.

TREATMENT

Caregivers prescribe a number of medications. These help control emotional and movement problems associated with HD. Most drugs used to treat the symptoms of HD have side effects. They include:

- Fatigue.
- Restlessness.
- Hyperexcitability.

It is important for people with HD to maintain physical fitness as much as possible. Individuals who exercise and keep active tend to do better than those who do not.

At this time, there is no way to stop or reverse the course of HD. Now that the HD gene has been located, investigators continue to study it. They want to understand how it cause disease in the human body.

RESEARCH BEING DONE

Scientific investigations use electronic and other technologies. They help scientists to see:

- What the defective gene does to various structures in the brain.
- How it affects the body's chemistry and metabolism.

Laboratory animals are being bred in the hope of duplicating the clinical features of HD. Then researchers can

learn more about the symptoms and progression of HD. Investigators are implanting fetal tissue in rodents and nonhuman primates. They hope to understand, restore, or replace functions typically lost by neuronal degeneration in individuals with HD. Related areas of investigation include:

- Excitotoxicity (overstimulation of cells by natural chemicals found in the brain).
- Defective energy metabolism (a defect in the mitochondria).
- Oxidative stress (normal metabolic activity in the brain that produces toxic compounds called free radicals).
- Tropic factors (natural chemical substances found in the human body that may protect against cell death).

Research is continually being done to learn more about this problem.
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