Participating in Research

Clinical studies and trials are how new knowledge is generated, and vital to advancing scientific discoveries in HD/JHD. Being a research participant is not for everyone, but without participants there can not be new treatments in the future.

The following links will give you additional information on research participation:

hsglimited.org

clinicaltrials.gov

hdsa.org/hd-research/hd-trial-finder

Additional Web Based Resources

Help 4 HD International
help4hd.org

Huntington Study Group (HSG)
huntingtonstudygroup.org
Factor-H
factor-h.org
HD GEM
hdgem.org
Social Security
ssa.gov/compassionateallowances/
National Organization for Rare Disorders (NORD)
rarediseases.org
HD Reach
HDReach.org
Huntington's Disease Youth Organization (HDYO)
HDYO.org
HOPES (Stanford)
hopes.stanford.edu
National Alliance on Mental Health (NAMI)
NAMI.org
National Institute of Health (NIH)
NIH.gov
National Suicide Prevention
suicidepreventionlifeline.org
Champions for HD
championsforhd.org
Meg's Fight For a Cure:
megsfightforacurejhd.com
chANGE-HD
Medicine.uiowa.edu/psychiatry/research/change-hd
HD Buzz
en.hdbuzz.net

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Help 4 HD International

Website: www.Help4HD.org
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What is HD?

- What is Huntington's Disease?
- Common symptoms of HD
- Treatments and Medications
- Testing and Family Planning
- Participating in Research
- Additional Web Based Resources

Help 4HD

Help 4 HD International

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What is Huntington's Disease

Huntington's disease (HD) is an inherited disorder that causes brain cells, called neurons, to die in various areas of the brain, including those that help to control voluntary (intentional) movement. Symptoms of the disease, which gets progressively worse, include uncontrolled movements (called chorea), abnormal body postures, and changes in behavior, emotion, judgment, and cognition. People with HD also develop impaired coordination, slurred speech, and difficulty feeding and swallowing. HD typically begins between ages 30 and 50. An earlier onset form called juvenile HD occurs under age 20. Its symptoms differ somewhat from adult onset HD and include rigidity, slowness, difficulty at school, rapid involuntary muscle jerks called myoclonus, and seizures. More than 30,000 Americans have HD.

Huntington's disease is caused by a mutation in the gene for a protein called huntingtin. The defect causes the cytosine, adenine, and guanine (CAG) building blocks of DNA to repeat many more times than is normal. Each child of a parent with HD has a 50-50 chance of inheriting the HD gene. A child who does not inherit the HD gene will not develop the disease and generally cannot pass it to subsequent generations. A person who inherits the HD gene will eventually develop the disease. HD is generally diagnosed based on a genetic test, medical history, brain imaging, and neurological and laboratory tests. ***From the NIH's website 2021***

Common Symptoms of HD

Each person with HD experiences a different combination of symptoms. Here are some of the most common:

Cognitive decline

Chorea (involuntary body/facial movements)

Hyper/hypo sexuality

Behavioral issues including rage

Impulsivity/perseveration

Gastrointestinal/urological changes

Gait instability

Sleep disturbances

Impaired swallowing/aspiration

Memory loss, especially short-term

Auditory and/or visual hallucinations

Delayed responses

Lack of awareness

Mood disorders such as depression, anxiety, and compulsiveness

Treatments and Medications

As of 2021, there are only two drugs approved by the FDA for treatment in Huntington's Disease. These medications are Austedo and Tetrabenazine and are used to treat chorea. There are many medications developed for other disorders that are used successfully to treat additional symptoms of HD. Clinical trials are ongoing in the development of new symptomatic treatments. Currently, no treatment exists that delays the onset or slows the progression of the disease.

Testing and Family Planning

Predictive Testing: Since the gene that causes Huntington's disease was found in 1993, people at risk over 18 years of age can undergo predictive testing. Genetic counseling is highly recommended for people considering predictive testing. It is not recommended to test minors (under 18) unless they are showing symptoms.

Confirmatory Testing: Diagnosis of Huntington's disease is made based on a neurological exam, family history, and confirmatory genetic testing.

Family Planning

People who have Huntington's disease have a 50% chance of passing the gene on to their offspring. Since this is a genetic disease, family planning options include:

Natural Conception

Adoption

Sperm or Egg Donation/Surrogate

IVF PDG: In Vitro Fertilization (IVF) is an assisted fertility medical procedure where an egg is fertilized by the sperm outside of the body, and implanted into the uterus. Preimplantation genetic diagnosis (PGD) can be performed on the fertilized egg prior to implantation. Only eggs that do not show the HD gene expansion would be implanted.

Prenatal diagnosis: Doctors can perform a prenatal diagnosis, where the fetus is tested for HD.