What is Huntington's Disease?

Huntington disease is a neurodegenerative genetic progressive brain disorder that causes uncontrolled movements, emotional problems, and loss of thinking ability (cognition), according to the NIH (National Institutes of Health). Mutations in the HTT gene cause Huntington disease. The HTT gene provides instructions for making a protein called huntingtin.

However, Huntington disease is much more complicated than this description. In this brochure we will touch on the very basics of what Huntington's disease is and what is available for its support, treatment and care. Although there is no cure for HD at this time, there is much hope through accelerated scientific research, regenerative medicine, antisense drug technology and clinical drug trials underway.

What is Juvenile Huntington's Disease?

Juvenile Huntington Disease is a more virulent form of HD and starts in childhood or adolescence. Only about 5% of HD cases are JHD, but the symptoms and progression are horrible and devastating. Onset may present with decline in scholastics, behavior, balance, and speech.

NIH states that seizures occur in 30% to 50% of JHD children and progresses much more quickly than adult Huntington disease.

According to Dr. Peg Nopoulos of the University of Iowa, Kids-HD/Kids-JHD, a natural history study sponsored by CHDI, JHD is diagnosed when their CAG is over 60 and before the age of 21 years.

HD: A Trinucleotide Disorder

The HTT mutation that causes Huntington disease involves a DNA segment known as a CAG trinucleotide repeat. This segment is made up of a series of three DNA building blocks (cytosine, adenine, and guanine) that appear multiple times in a row. Normally, the CAG segment is repeated 10 to 35 times within the gene. In people with Huntington disease, the CAG segment is repeated 36 to more than 120 times. People with 36 to 39 CAG repeats may or may not develop the signs and symptoms of Huntington disease, while people with 40 or more repeats almost always develop the disorder. (Information referenced from NIH.gov)

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Care Centers for HD

There are many neurology and movement disorder clinics and centers throughout the US that specialize in Huntington disease, Parkinson disease and other movement disorders. The HDSA (Huntington's Disease Society of America) supports a number of Centers of Excellence in the US. Link: http://hdsa.org/about-hdsa/centers-of-excellence/. Most University Hospitals and Clinics will have an excellent Neurology Department or Movement Disorder Clinic. Ask your primary physician for a referral to the nearest expert clinic.

Clinical Trial Opportunities

Clinical studies and trials are vital to advancing scientific research to discover treatments, therapies and possibly the cure. The benefit of participating in trials is that the HD individual will be contributing to the advancement of science and they will also be able to get expert care from an experienced Huntington's multidisciplinary team. It's a fabulous way to link up with the best of the best. Here are some clinical trial information links:

- EHDN Pride-HD: www.euro-hd.net (Europe)
- Enroll HD: www.enroll-hd.org
- HDSA: www.HDTrialFinder.org
- HSG (Huntington Study Group): www.hsglimited.org

Diagnosing HD and JHD

Diagnosis may be made at any time during a person's lifetime with neurological exam, family and personal medical history and genetic blood test. Genetic counseling is highly recommended, as the diagnosis of Huntington disease or Juvenile Huntington disease is very devastating and there is a high suicide rate among diagnosed individuals (about 7%).

Discovering the Gene

Thanks to the HDF, the ongoing US-Venezuela Huntington's Disease Collaborative Research Project was started in 1979, and reported a major breakthrough in 1983 with the discovery of the approximate location of a causal gene. In 1993, the research group (HDFoundation.org) isolated the precise causal gene at 4p16.3, making this the first autosomal disease locus found using genetic linkage analysis.

Options for Prevention

Since there is no cure, treatment or therapy to stop progression, the only way to prevent HD is through IVF PGD (Invetro Fertilization with Preimplantation Genetic Diagnosis). PGD is a procedure in which one of the cells (blastomere) of the embryo is removed for genetic analysis three days after egg retrieval to identify genetically abnormal embryos.

Research for HD

*These are just a few locations for HD and *JHD research:

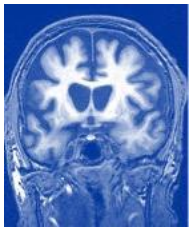
CHDI	Mass General	UCR
CIRM	Georgetown	UCSF
Gladstone	NINDS	UCSF MAC
Harvard	Stanford	UCSD
HDF	*UC Davis	*UI
HDSA	UCI MIND	UR
IU	UCLA	VCU

Resources

H4HDiRegister.org
HDCare.org (UCI)
HDDrugWorks.org
HDFoundation.org
HDLF.org
HDSA.org
HDReach.org
HDYO.org
Help4HD.org
Help 4 JHD
Help4HD Radio/BTR
HelpforHDFamiliesNFP.org
HOPES (Stanford)
Kids-HD/Kids-JHD (UI)
NAMI.org
NIH.gov
Research4HD.org
SuicidePreventionLife-Line.org
The Huntington's Post

SSA CALs

As of year end 2012, the Social Security Administration added Juvenile Onset and Adult Onset Huntington Disease to its Compassionate Allowance List which is designed to fast track the disability process for individuals. However, the 24 month waiting period for Medicare benefits is still in effect.



FROM WIKIPEDIA: Coronal section from a MR brain scan of a patient with HD, showing atrophy of the heads of the caudate nuclei, enlargement of the frontal horns of the lateral ventricles (hydrocephalus*ex vacuo*), and generalized cortical atrophy.

Symptoms

- Anososgnosia (lack of awareness)
- Chorea (uncontrolled dance -like movements)
- Delayed response
- Dystonia (more common in JHD)
- Dysphagia
- Hallucinations
- Memory loss
- OCBs
- Perseveration (repetitive words/thoughts/actions)
- Poor judgment
- Psychiatric behaviors
- Rages
- Rigidity
- Seizures (more common in JHD)
- Stumbling gate

Testing Labs

In general, there is an inverse relationship between the number of repeats and the severity of disease; that is, the larger the repeat size, the more severe the symptoms and the earlier the onset of disease. In addition to this, mutated alleles are genetically unstable and have a tendency to undergo further expansion as they are transmitted to future generations, increasing the disease severity in subsequent generations. This phenomenon is known as anticipation (according to labtestonline.org).

Treatments

Dr. LaVonne Goodman from HDDrugWorks.org has developed a set of algorithms through surveys of Huntington disease medical and psychiatric professionals throughout the US and Europe for the treatment of some common symptoms of HD: Obsessive Compulsive Behaviors, Irritability, and Chorea. You may find these algorithms at:

www.hddrugworks.org