



Fact Sheet: Huntington's Disease

Definition

Huntington's disease is a hereditary disorder of the central nervous system. Symptoms typically appear in adult life between the ages of 30 and 50. It is most often characterized by chronic progressive chorea and dementia without remissions.

Facts

It is estimated that 30,000 persons have Huntington's disease nationwide. Another 150,000 are at a 50 percent risk of inheriting the disease from an affected parent. The age of onset can range from age two to over age 80. The disease runs a relentless degenerative course over a period of 10-25 years. The affected person eventually succumbs to complications such as heart failure or aspiration pneumonia.

Symptoms

An individual with Huntington's disease often has problems in three areas: control of bodily movements, intellectual functioning and emotional control. The intensity and number of the above symptoms, however, varies with each Huntington's disease patient.

Chorea—quick, jerking, uncontrollable movements of the limbs, trunk and face are the most

obvious symptoms. Movements tend to become larger over time. The person's gait is often mistaken for that of a person who is drunk. Rigidity or muscle stiffness may occur later on. Speech and swallowing are typically affected. In the late stages of the disease, choking on food can be a major concern.

Poor short-term memory and judgment—Minor intellectual changes are often the first sign of cognitive impairment. Difficulty organizing routine matters or coping with new situations may occur. Verbal fluency may be affected. Although most affected individuals continue to recognize their caregivers and can give feedback about their condition and care, the person with Huntington's disease suffers declining mental skills as the disease progresses.

Depression, irritability and apathy can be noted before and after the onset of chorea in a percentage of the Huntington's disease population. Some individuals may exhibit schizophrenic-like behavior, such as hallucinations, mania, anxiety and psychosis.

Diagnosis of Huntington's Disease

If there is a positive family history of Huntington's disease, the presence of an

extrapyramidal movement disorder (when nerve impulses are not properly transferred to

the muscles) is usually enough to confirm a diagnosis. Conventional neurological examinations and the presence of depression and other psychological disturbances are not enough to provide an accurate diagnosis.

The Huntington's Disease Gene and Genetic Testing

In 1993 scientists announced the discovery of the gene which causes Huntington's disease. This breakthrough has led to the development of a direct gene test for HD which can be used to predict the onset of Huntington's before symptoms develop.

The gene discovery shows that Huntington's disease is caused by a "genetic stutter"-a stretch of DNA repeated over and over at one end of a gene on Chromosome four. Efforts continue to determine precisely how this genetic mutation leads to the symptoms of the disease.

The test itself is a blood test. However, a positive test result will not indicate when symptoms will appear. Presymptomatic testing should include genetic counseling, a neurologic exam, a psychological interview, discussion of results and a follow-up. In the absence of a cure, at-risk individuals may prefer to live with uncertainty, rather than take the test. The decision must be a personal one and should be discussed with a genetic counselor.

Contact the Huntington's Disease Society of America (HDSA) for a list of testing centers (see RESOURCES section).

Research

Recent research findings revealed that cell death in Huntington's disease may be caused by a ball of protein that forms in the nucleus of the cell. Current research is exploring possible

drug treatments which would prevent the accumulation of protein in cells.

Other research efforts include the development of a mouse model for Huntington's disease and the CARE-HD study, a clinical drug trial underway at about 20 Huntington Study Group sites. Here researchers are evaluating the combination of a medication (remacimide) and co-enzyme Q-10. Both basic (laboratory) and clinical (testing of medications and treatments) research continue to pursue avenues to facilitate new drug testing and experimental surgical techniques.

Treatment

Medication can help the involuntary movements and emotional disorders for some Huntington's disease patients. Because of the strong emotional impact of the diagnosis and the stress of living with the knowledge that the children in the family may be at risk, family counseling and/or participation in support groups can be very helpful.

Today, more than ever, there is hope for people with Huntington's disease. Rapid progress is being made toward potential treatments for Huntington's disease. Likewise, quality of care for people with Huntington's disease has improved greatly in recent years. By seeking early intervention from health professionals, such as physical, occupational and speech-language therapists, the person with Huntington's disease can have more control over the disease and over his/her life. By learning strategies to help them cope with the many changes that are brought on by Huntington's disease, people can live meaningful, productive lives well into the final stages.

Recommended Reading

Comprehensive Curriculum for Care Providers to the Huntington's Disease Affected Community (CD-ROM) 1997, a joint project of Family Caregiver Alliance and the Huntington's Disease Association of America. Available from FCA.

Genes & Generations: Living With Huntington's Disease, Alison Gray, 1995, Wellington Huntington's Disease Association, Social Work Dept. (Res. 2), Wellington Hospital, Private Bag 7902. Wellington South, New Zealand. To order, send \$15.00 check payable to Wellington Huntington's Association.

A Physician's Guide to the Management of Huntington's Disease: Pharmacologic and Non-Pharmacologic Interventions, Neal Ranen, Carol Peyser, Susan Folstein, 1993, Huntington's Disease Society of America (HDSA), New York, NY.

Living With Juvenile Huntington's Disease, edited by Wendy Elliott, 1993, Huntington Society of Canada, Cambridge, Ontario. Available from HDSA.

Contact HDSA to obtain a current publications list which includes written and audiovisuals pertaining to all aspects of Huntington's disease.

Credits

Huntington's Disease Society of America, Genetic Testing for Huntington Disease, 1996.

Scientists Report New Understanding of Cell Death in Huntington's Disease, The Marker, Vol. 10(2), Summer 1997, HDSA.

Dubinsky, Richard, M.D., Huntington's Disease, Kansas University Medical Center, Dept. of Neurology, Kansas City, KS, October 1997.

Resources

Mountain Caregiver Resource Center
2491 Carmichael Drive, Suite 400
Chico, CA 95928
(530) 898-5925
(800) 995-0878
www.caregiverresources.org

Mountain Caregiver Resource Center offers a full complement of family services designed to assist caregivers of adults with brain disorders. Most services are offered free or on a low-cost sliding scale.

Services include: specialized information, family consultation and care planning, support groups, short-term counseling, respite care, legal and financial assistance, in addition to family and professional training.

Family Caregiver Alliance
690 Market Street, Suite 600
San Francisco, CA 94104
(415) 434-3388
(800) 445-8106 (in CA)
Website: <http://www.caregiver.org>
E-mail: info@caregiver.org

Family Caregiver Alliance supports and assists caregivers of brain-impaired adults through education, research, services and advocacy.

FCA's information Clearinghouse covers current medical, social, public policy and caregiving issues related to brain impairments.

For residents of the greater San Francisco Bay Area, FCA provides direct family support services for caregivers of those with Alzheimer's disease, Huntington's disease, stroke, head injury, Parkinson's and other debilitating brain disorders that strike adults.

Huntington's Disease Society of America
158 West 29th Street, 7th Floor
New York, N.Y. 10001-5300
(212) 242-1968
(800) 345-HDSA

The Huntington's Disease Society of America (HDSA) supports research on Huntington's disease, produces and disseminates educational materials for the public and professionals, and provides services for people with Huntington's disease and their families.

Regional HDSA chapters across the U.S. provide information and referrals, operate education and publicity programs, run support groups, and offer a range of services for people with Huntington's disease and their families. California chapters of HDSA are listed below:

Greater Los Angeles Chapter
9903 Santa Monica Blvd., Ste. 106
Beverly Hills, CA 90212
(213) 294-7250

Northern California Chapter
1212 Broadway St., Ste. 830
Oakland, CA 94612
(916) 962-2436 (Helpline)
(888) 828-7348 (toll free in CA)

San Diego County Chapter
P.O. Box 152263
San Diego, CA 92195-2263
(619) 552-8585 ext. 3431

National Society of Genetic Counselors
233 Canterbury Dr.
Wallingford, PA 19086-6617
(610) 872-7608

The National Society of Genetic Counselors (NSGC) can assist individuals and their families in locating a genetic counselor. NSGC does not, however, maintain information on specific genetic disorders.

Genetically Handicapped Persons Program
State of California Department of Health Services
714 "P" St., Room 300
Sacramento, CA 95814
(916) 654-0503
(800) 639-0957

The GHPP is a State program which provides coordination of care and helps pay for medical costs of persons with Huntington's disease and other disorders.

Reviewed by the Huntington's Disease Society of America. Prepared by Family Caregiver Alliance in cooperation with California's Caregiver Resource Centers, a statewide system of resource centers serving families and caregivers of brain-impaired adults. Funded by the California Department of Mental Health. Revised December 1997. ©All rights reserved.

Revised 09/27/02