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Huntington's disease: a clinical review

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Abstract

Huntington disease (HD) is a rare neurodegenerative disorder of the central nervous system characterized by unwanted choreatic movements, behavioral and psychiatric disturbances and dementia. Prevalence in the Caucasian population is estimated at 1/10,000-1/20,000. Mean age at onset of symptoms is 30-50 years. In some cases symptoms start before the age of 20 years with behavior disturbances and learning difficulties at school (Juvenile Huntington's disease; JHD). The classic sign is chorea that gradually spreads to all muscles. All psychomotor processes become severely retarded. Patients experience psychiatric symptoms and cognitive decline. HD is an autosomal dominant inherited disease caused by an elongated CAG repeat (36 repeats or more) on the short arm of chromosome 4p16.3 in the Huntingtine gene. The longer the CAG repeat, the earlier the onset of disease. In cases of JHD the repeat often exceeds 55. Diagnosis is based on clinical symptoms and signs in an individual with a parent with proven HD, and is confirmed by DNA determination. Pre-manifest diagnosis should only be performed by multidisciplinary teams in healthy at-risk adult individuals who want to know whether they carry the mutation or not. Differential diagnoses include other causes of chorea including general internal disorders or iatrogenic disorders. Phenocopies (clinically diagnosed cases of HD without the genetic mutation) are observed. Prenatal diagnosis is possible by chorionic villus sampling or amniocentesis. Preimplantation diagnosis with in vitro fertilization is offered in several countries. There is no cure. Management should be multidisciplinary and is based on treating symptoms with a view to improving quality of life. Chorea is treated with dopamine receptor blocking or depleting agents. Medication and non-medical care for depression and aggressive behavior may be required. The progression of the disease leads to a complete dependency in daily life, which results in patients requiring full-time care, and finally death. The most common cause of death is pneumonia, followed by suicide.