

# Chapter 13

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## Huntington Disease

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## What are the features and diagnosis of cognitive symptoms in Huntington disease?

### Answer

Among the cognitive impairments in Huntington's disease, memory function and executive function are particularly affected. Psychiatric symptoms such as depression, anxiety, irritability, apathy, and perseveration are often observed. The definitive diagnosis is by genetic diagnosis. B

### Comments and evidence

Huntington's disease is an autosomal dominant hereditary disease, manifesting adult-onset chorea and psychiatric symptoms as the main symptoms. The causative gene of Huntington's disease is the huntingtin (*HTT*) gene located in the short arm of chromosome 4 (4q16.3). The gene consists of an unstable repeated CAG sequence, and abnormal expansion of this sequence leads to the development of Huntington's disease. While the number of CAG repeats in a normal allele is less than 26, an increase in number of repeats to over 36 is considered to cause Huntington's disease. In addition, the onset age becomes younger and the disease becomes more severe in successive generations (genetic anticipation), and inheritance from the father may result in more severe disease. Core dementia symptoms are personality changes and cognitive impairment. On the other hand, affective symptoms such as emotional lability, losing temper, irritability, stubbornness and apathy are present to various degrees<sup>1)</sup>. The definitive diagnosis is by genetic testing<sup>2, 3)</sup>, and this genetic test (*HTT* gene CAG repeat sequence analysis) is covered by health insurance in Japan.

### ■ References

- 1) Shannon KM. Huntington's disease-clinical signs, symptoms, presymptomatic diagnosis, and diagnosis. *Handb Clin Neurol.* 2011; 100: 3-13.
- 2) Ross CA, Aylward EH, Wild EJ, et al. Huntington disease: natural history, biomarkers and prospects for therapeutics. *Nat Rev Neurol.* 2014;10(4): 204-216.
- 3) Reilmann R, Leavitt BR, Ross CA, et al. Diagnostic criteria for Huntington's disease based on natural history. *Mov Disord.* 2014; 29(11): 1335-1341.

### ■ Search formula

PubMed search: May 28, 2015 (Thursday)  
#1 "Huntington Disease/diagnosis" [Mesh]

Ichushi search: May 28, 2015 (Thursday)  
#1 (Huntington disease OR "Huntington Disease") AND ((SH = Diagnostic use, diagnosis, diagnostic imaging, X ray diagnosis, radionuclide diagnosis, ultrasound diagnosis) OR Symptom OR Feature OR Syndrome OR Sign OR Finding)