How can progression of Huntington’s disease (HD) be measured?

HD is a multidimensional disease, with patients experiencing a wide range of symptoms that negatively impact their overall quality of life. Subtle symptoms can be present for many years before a diagnosis of HD is made.

Stages of HD:

1. Pre-symptomatic
   HD is a genetic (inherited) disease, but patients don’t initially experience symptoms. The mean age of onset for diagnosis of motor symptoms (affecting movement) is 45 years.

2. Prodromal
   Gradual appearance of subtle signs and symptoms of HD.

3. Early
   Increased symptoms begin to affect ability to carry out daily tasks, motor diagnosis received.

4. Moderate
   Worsening of symptoms leading to a dramatic reduction in quality of life.

5. Advanced
   Very severe impairment of movement and profound physical disability. Dementia-like symptoms.

In patient registries (e.g. Enroll-HD) and clinical trials, doctors and scientists use assessment scales to diagnose and monitor the progression of these symptoms.

Symptoms usually affect three main areas or domains: movement, cognitive (difficulties with memory, thinking and planning) and psychiatric (changes in behaviour and personality).

Cognition
- Ability to think clearly and quickly
- Attention
- Perception of time
- Difficulty learning new tasks

Motor function
- Jerky, involuntary movements
- Balance issues
- Fine motor issues
- Speech and swallowing issues

Behaviour
- Depression
- Apathy
- Irritability and angry outbursts
- Anxiety

Other/impacts
- Sleep problems
- Inability to work
- Isolation
- Pain

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Rating scales for HD typically measure individual clinical domains or they summarise a number of assessments, which may have limited sensitivity for measuring changes associated with disease progression\(^6,7\)

The Unified Huntington’s Disease Rating Scale (UHDRS) is a collection of assessments of clinical HD features, including motor function, cognition, behaviour, and overall function\(^8\)

**Motor assessment**
- Total Motor Score (TMS)

**Cognitive assessment**
- Symbol Digit Modalities Test (SDMT)
- Stroop Word Reading (SWR)

**Independence scale**
- Total Functional Capacity (TFC) scale

**Overall function**
- Total Functional Capacity (TFC)

**Motor function**
- Total Motor Score (TMS)

**Behavioural assessment**
- Stroop Word Reading (SWR)

**Cognitive function**
- Symbol Digit Modalities Test (SDMT)

Studies using patient registry data (TRACK-HD, CARE-HD, COHORT, 2CARE) show that the cUHDRS provides an improved measure of clinical progression relative to individual motor, cognitive and functional outcome measures, in terms of both sensitivity and association with measures of brain atrophy\(^7\)

The cUHDRS is valid, reliable, able to detect change, and associated with clinically meaningful differences (both cross-sectional and longitudinal) in function and independence across the spectrum of patients with early-manifest HD\(^9\)