NEUROPSYCHOLOGICAL ASSESSMENT OF A PATIENT DIAGNOSED WITH MAJOR DEPRESSION AND HUNTINGTON'S DISEASE

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INTRODUCTION

Huntington's disease (HD) is a chronic progressive neurodegenerative disorder associated with corticostriatal pathology caused by an extension of a part of a gene leading to extensive nerve cell loss. The more times the expansion section recurs, the earlier the disease begins. HD is inherited in an autosomal dominant pattern, and thus, requires only one parent for a 50% chance to inherit the abnormal gene. Diagnosis is confirmed with genetic testing. The disease affects mobility, cognitive functions, behavior and personality. It typically starts between the ages of 30 and 50, but there are cases of patients with onset of the disorder at a younger age. Medication targets to alleviate psychiatric and mobility symptoms, but the disorder itself is not curable (Nopoulos 2016).

In the early stages, HD is difficult to diagnose, especially if the family history is unknown, as the symptoms are complex and variable from patient to patient. The most common clinical symptomatology involves uncontrolled movements resembling dancing moves, called chorea, as well as difficulty speaking and walking. Initially, symptoms such as depression may be more pronounced than chorea. Mild mental, emotional, and behavioral changes may also occur before the clinical symptoms (memory impairment, poor concentration, difficulty performing scheduled tasks, impulse control disorder, depression, sleep disorders, sexual problems, difficulty swallowing and loss of balance with contact).

As the disorder progresses, involuntary movements increase, more serious deficits in thought content, learning and memory occur, emotional and behavioral changes intensify, as well as disorientation and difficulty in speaking and swallowing. In subsequent stages of the disease, patients are able to understand the daily routine and recognize other people, but face difficulty in taking self-care, bladder incontinence, lose the ability to walk, and therefore require support from a caregiver. The disease leads to life-threatening conditions due to cardiopulmonary complications (pneumonia and cardiovascular diseases are the most frequent primary causes of death)(Nopoulos 2016).

HD Cognitive deficits

Attention

As time and HD progresses, the range of attention usually seen with immediate recall, declines. Concentration and mental visual cognition are affected at all stages of the disease. Attention may be normal in the early stages of the disease, but shortens abruptly in the following stages, along with difficulty in maintaining and relocating attention (Paulsen 2011, Snowden 2017).

Memory

Several studies have described specific patterns of memory deficits among HD patients; mild in the early stages, but progressively worsening with greater intensity and severity in memory span (Snowden 2017). The characteristics of this pattern include impaired short-term memory which is extremely vulnerable to intrusive effects. Learning new information material decelerates (storage deficit). This problem is exacerbated by retrieval deficit, and thus, it becomes evident especially in recall tests. Semantic guidance or recognition formation improves retrieval (Van Etten et al. 2019).

Unlike normal individuals, HD patients cannot spontaneously utilize learning strategies, such as rehearsal or coding through mental imagery. Serial learning becomes virtually impossible, as in story retrieval, as partial loss of information occurs after a period of delay. Nevertheless an emotionally charged material can have a reinforcing effect and the reinforcement seems to be retained in the delayed recall. These patients are aware of their memory impairment (Cleret de Langavant et al. 2013, Lezak et al. 2009).

Language

The structure of language, vocabulary, grammar and syntax tends to persist until the final stages, where dementia becomes virtually universal. However, verbal

production becomes simplified, shortened, and prone to semantic errors. As motor and cognitive symptoms deteriorate, patients eventually stop talking altogether, due to the very loss of voluntary control of speech and breathing muscles, rendering both food intake and swallowing difficult and dangerous (Lezak et al. 2009).

Spatial function deficits

Almost all studies report impairment of visuospatial ability, including right-left orientation, regardless of whether motor response is required (Snowden 2017). Visual construction and path learning can, however, be intact in mildly disturbed patients (Lezak et al. 2009, Salmon & Bondi 2009, Smith & Bondi 2013).

Executive functions

Executive deficiencies are similar to those seen in patients with frontal lobe lesions and include reduced self-producing activity, impaired behavioral regulation, and deficits in planning and organization (Bonelli & Cummings 2007, Sitek et al. 2014).

CASE STUDY

A 29-year-old female patient, university graduate and private employee, with a two-year individual HD history, was examined on a scheduled appointment. During history-taking, the patient presented herself as a person of moderate sociability with difficulty making interpersonal relationships. No cases of conduct disorders or suicidal ideation were reported, but the patient's attitude indicated emotional instability with intense anxiety and bursts of tears, during history-taking. During the most recent four months, the patient self-reported the occurrence of episodes of psychomotor anxiety with depressive symptoms for which no medication was ever followed.

From the patient's history in the first year neither the necessity to go beyond the basic set of assessment of cognitive abilities arose nor the addition of specialized tests.

The patient's follow-up neuropsychological examination focused on memory function (story memory, reverse number retrieval, verbal flow test), attention (Stroop, direct number retrieval, TMT part A), and executive functions (Trail Maiking Test part B, cube test), Anoi tower, Test of opto-spatial functions). Her score on both immediate and long-term memory tests (story memory, reverse number retrieval, verbal flow test) appeared deficient for the patient's age and cognitive level. Regarding the long-term retention at the specific time of the evaluation, difficulties were observed in the

tests that allow the conceptual organization of the data to be memorized (story memory). The maintenance of mnemonic traces concerning general knowledge (declarative memory) fluctuated at normal levels. The patient's attention span fluctuated at very low levels as indicated by the slow execution of the AD part of the Trail Making Test but also by its performance in the Stroop interpolation condition. In general, the patient's performance in information processing speed, learning, memory and executive function ranged below the normal limits for her age and level of education at that time. In particular, the patient showed reduced accuracy and speed of rotation of visual attention to a different type of reaction, difficulty in abstract thinking and parallel processing of information according to its performance in the individual tests of the neuropsychological array. The patient at the time of the neuropsychological assessment met the criteria of moderate cognitive impairment with the presence of deficits in the dimensions of attention (ability to maintain a stable behavioral response), vigilance (ability to respond), and general alertness (in stimuli and readiness for response). It showed deficits in the dimensions of both immediate and long-term memory. However, it showed that it maintains at normal levels the ability to perform daily activities as well as to assess/control reality. The values in the STAI test were indicative of intense stress (condition and structure) and the BDI value was characteristic of major depression (Table 1). The values in the EPQ test were indicative of introversion, pessimism and tendency towards social isolation. While in the SCL 90-R test the index of annoyance from positive symptoms and six scales showed pathological values (somatization, compulsion, depression, anxiety, anger, paranoid ideation and psychosis).

CONCLUSIONS

Given the patient's HD diagnosis, the extent of physical, mental and cognitive disorders as well as the high levels of anxiety and depression are partially justified. There was also a fairly large range of cognitive deficits in this patient, the progression of which, however, could not be determined before its reassessment after at least one year. Particularly affected were its executive functions which involved design, cognitive flexibility, abstract thinking, rule acquisition, information processing speed, and the initiation of appropriate actions. The main mnemonic deficits concerned her short-term memory. The pattern of her cognitive deficits combined with her medical history and neurological and psychiatric assessment led to the diagnosis of onset HD cortical dementia syndrome.

Table 1. Scores for STAI and BDI assessment

Inventory	Scoring	Mean Value	S.	Normal Value Range (for 1 S.)
STAI State	57	24.95	11.36	13.59–36.31
Trait	63	27.88	11.43	16.45–39.31
BDI	27	Depression (severe)		

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