**Scholars Journal of Medical Case Reports**
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

**Psychiatry**

# When Psychiatric Symptoms Reveal Huntington's disease: An Illustrative Case Study

A. K. Rhaouti[1*], S. Stati[1], A. Khallouk[1], H. Boukidi[1], A. Boukdir[1], O. Belakbir[1], M. Sabir[1]

[1]Psychiatry Resident, Ar-Razi Psychiatric University Hospital of Salé

**DOI:** https://doi.org/10.36347/sjmcr.2025.v13i01.022 | **Received:** 13.09.2024 | **Accepted:** 18.10.2024 | **Published:** 15.01.2025

***Corresponding author:** A. K. Rhaouti
Psychiatry Resident, Ar-Razi Psychiatric University Hospital of Salé

**Abstract** **Case Report**

***Introduction*:** Huntington's disease (HD) is a rare genetic disorder with autosomal dominant inheritance, caused by CAG repeat expansions in the IT15 gene. It affects about 1 in 10,000 people and leads to neurodegeneration, primarily impacting the striatum and later causing cortical atrophy. Clinically, HD presents with motor, cognitive, and psychiatric symptoms, with psychiatric manifestations often occurring early and causing diagnostic confusion. ***Clinical Observation*:** A case of Mrs. S.M., a 40-year-old woman hospitalized for aggressive behavior, is discussed. Initially diagnosed with brief psychotic disorder, she later developed dysarthria and choreic movements. Genetic testing confirmed HD, underscoring the complex psychiatric presentation that can precede motor symptoms. ***Discussion*:** Psychiatric symptoms are common in HD and may appear before motor issues, often leading to misdiagnosis. These symptoms, including aggression, depression, and psychosis, correlate with underlying brain degeneration. Treatment generally involves atypical antipsychotics like quetiapine for better tolerance. ***Conclusion*:** HD's early psychiatric symptoms can obscure diagnosis. Effective management focuses on multidisciplinary care to sustain patient autonomy and quality of life.

**Keywords**: Huntington's Disease(HD), Genetic Disorder, Huntingtin Gene, Basal Ganglia, Striatum.



## INTRODUCTION

Huntington's disease (HD) is a rare genetic disorder transmitted in an autosomal dominant manner and characterized by complete penetrance, meaning that an individual carrying the defective gene will develop the disease at some point in their life [1]. It results from the pathological amplification of CAG (cytosine-adenine-guanine) repeats in the IT15 gene, which codes for huntingtin, a protein whose function remains largely unknown [1]. The prevalence of this disease is relatively low, affecting approximately 1 in 10,000 people [2].

This neurodegenerative condition primarily affects the striatum, particularly the caudate nucleus and putamen, but ultimately leads to atrophy of the cerebral cortex and basal ganglia [2]. Clinically, HD manifests as a triad of symptoms: chorea-like motor disorders, progressive cognitive deficits, and psychiatric symptoms. The latter, which include behavioral changes, irritability, mood disorders, and sometimes hallucinations, can constitute the initial signs of the disease in 20 to 80% of cases, frequently leading to diagnostic errors [3].

This work aims to describe the initial psychiatric manifestations of HD through the analysis of a representative clinical case. This study highlights the diagnostic challenges due to the diversity and nature of the early psychiatric symptoms associated with HD, which can easily be confused with other psychiatric disorders [3].

## CLINICAL OBSERVATION

The case involves Ms. S.M., a 40-year-old illiterate woman, hospitalized in October 2023, in the "A" psychiatry department at Ar-Razi Salé Psychiatric University Hospital (Morocco), under compulsory hospitalization at the request of a third party due to hetero-aggressiveness. She is single. In her family history, a brother is followed for schizophrenia. In her personal history, she is monitored for stabilized type 2 diabetes under a diet alone. A week prior to her psychiatric admission, the patient reportedly exhibited a sudden change in behavior. She became aggressive, unstable, and very irritable, refusing any food due to fear of poisoning. One day before her hospitalization, she allegedly attempted to assault her cousin, causing several

bruises, which led to her compulsory psychiatric hospitalization.

During the psychiatric examination, Ms. S.M. was very unstable, had difficult contact with concentration and memory disturbances. Her speech was not spontaneous, conveying delusional ideas of persecution and witchcraft. She reported hearing threatening voices all day and seeing strange people around her. The somatic examination, biological assessments, and brain scan were normal.

The diagnosis of brief psychotic disorder was retained. Upon admission, Ms. S.M. was started on haloperidol at a dose of 10 mg per day: a classical antipsychotic (AP) chosen for its injectable form due to her refusal of treatment. After three weeks of hospitalization, there was a reduction in psychotic symptoms and behavioral disorders, allowing for Ms. S.M.'s discharge on haloperidol 2 mg/ml oral solution at a dosage of 100 drops per day. During the post-cure interview, she presented abnormal movements in the upper limbs as well as dysarthria, which did not improve with antiparkinsonian medications. She was referred to neurology for specialized management. In the context of the etiological assessment, biological and radiological explorations were normal.

Given the age of onset of the symptoms, the negativity of the assessment, and the presence of the triad (choreic movements, psychiatric disorders, and cognitive disturbances), the diagnosis of HD was suspected and then confirmed by genetic testing. The diagnosis of HD was announced by the neurologist, and the information was systematically given to the patient. Genetic counseling was proposed, and the patient was started on quetiapine at a dosage of 200 mg per day. The patient continued follow-up at outpatient psychiatry and neurology clinics, but in an irregular manner with poor treatment adherence.

## DISCUSSION

HD is most often described by its neurological component, namely chorea. However, psychiatric disorders are common, occurring in 35 to 75% of cases and present throughout the disease's evolution [3]. They can be initial in 20 to 80% of cases, potentially leading to misdiagnosis and delaying appropriate management [3]. Paulsen *et al*., [4], found that about 98% of patients exhibited at least one psychiatric symptom before the onset of motor symptoms of HD. This is also true for our patient, who presented with acute psychiatric disorders suggesting, initially, a diagnosis of brief psychotic disorder.

Some studies [5], have explored the link between the onset of HD and the presence of psychiatric disorders, but results are heterogeneous. This may partly be explained by the nonlinear evolution of psychiatric symptoms related to the differential degeneration of striato-cortical circuits [6].

Psychiatric symptoms can be grouped into three main categories: behavioral disorders, mood disorders, and psychotic disorders. Apathy, irritability, agitation, and hetero-aggressive gestures are predominant in HD, with prevalence rates ranging from 33 to 76% depending on the studies and stages of the disease [7]. These behavioral disorders are independent of the duration of the disease, the severity of chorea, and dementia. Aggression and irritability are generally related to the loss of frontal inhibition functions [7].

This was the case for Ms. S.M., who unexpectedly attempted to harm her cousin. Hypotheses explaining the increased occurrence of violent behaviors and medico-legal acts in neurological diseases, particularly in males, include elevated testosterone levels, neurophysiological dysfunctions affecting the frontal lobes and circuits involving the median hypothalamic nucleus, amygdala, and hippocampus, as well as neurochemical dysfunctions marked by decreased acetylcholine and serotonin levels [2]. Other studies have also emphasized the direct relationship between criminal acts and hypothalamic and limbic lesions, rather than environmental and familial factors to which these patients are also subjected [8].

Regarding mood disorders, depressive disorders are the most common, although bipolar disorders have also been described. Depression is the most prevalent psychiatric disorder associated with HD (39 to 53%) and may result directly from the degenerative process of the disease, rather than solely as a reactive modality to HD [11]. It can affect most patients at any stage of the disease [18]. Thus, vigilance should be maintained at all stages of HD, with systematic assessment of the severity of depression. Indeed, suicide can be observed at an early stage of the disease and in the presymptomatic stage, with a prevalence ranging from 8 to 20% according to studies [2]. From a psychopathological perspective, suicidal behavior in HD can partly be explained by the presence of psychiatric comorbidity, particularly depression [9], but the hereditary nature of HD complicates the psychopathological approach to self-aggressive behaviors. Additionally, HD itself constitutes a suicidal risk factor [10].

Various studies have focused on the association between HD and psychotic symptoms reminiscent of schizophrenia, proposing several hypotheses [11]. Some authors suggest that the psychotic symptoms occurring in HD patients result from the co-occurrence of the HD gene and a gene or group of genes "pro-schizophrenia." This raises the hypothesis that "pro-schizophrenia" genes may determine a form of subcortical dysfunction responsible for disorganized thinking and negative symptoms [12].

Indeed, psychosis in HD patients is typically associated with cognitive impairment and primarily occurs after the onset of motor symptoms [13]. Some studies have found a correlation between cognitive deficits and atrophy of the caudate nucleus and frontal cortex.

For the treatment of her psychotic disorder, Ms. S.M. was given classical AP treatment based on haloperidol, which was later replaced by quetiapine due to the worsening of her choreic movements. According to studies [14], atypical APs (quetiapine, risperidone, aripiprazole, olanzapine) are preferred for their better tolerance.

## CONCLUSION

Psychiatric disorders can herald HD, which can lead to misdiagnosis and delay adequate management. Each patient is a particular case due to the combination of their symptoms and the repercussions of their illness on familial, social, and professional levels. The objective of multidisciplinary management is to preserve the patient's autonomy, family harmony, professional life, and quality of life for as long as possible.

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