



## Huntingtons Disease: A Case Report

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### **Authors' contributions**

*This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.*

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**Case Study**

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## ABSTRACT

**Background:** Huntington's disease (HD) is a genetic neurodegenerative illness characterized by progressive nerve cell degeneration in the brain, mainly in the basal ganglia. It often manifests itself between the ages of 30 and 40. The disease is including inherited genetic genes, which means that the affected person inherits the gene from a parent who also has the same genes. In populations of western European origin, the incidence of inherited genetic diseases is 3-10 per 1,00000. In India, it is far less common.

**Case Presentation:** This is a case of a 57 -year-old female schoolteacher who was brought to our institution with a trembling movement all over the body and imbalancing while walking. The clinical presentation of characteristics such as difficulty controlling his hands and fingers due to involuntary, uncontrolled motions is used to make the diagnosis He walked without a cane and seemed to be in good physical shape, yet when asked to sit, he slumps heavily into the chair. CT or MRI scans indicate lacunar ischemic foci in bilateral periventricular white matter. Mild cerebral and cerebellar atrophy was present. Complete blood testing were also carried out.

**Conclusion:** There are currently few therapies available. The emergence of treatment methods capable of directly targeting HTT, on the other hand, signals a new phase in HD research. There is now more than ever a serious possibility of modifying and preventing HD.

**Keywords:** Genetic disorders; hereditary; huntington disease; huntington chorea; neurodegenerative; case report.

## 1. INTRODUCTION

Huntington's disease is a hereditary nervous system disorder that produces uncontrolled choreiform movement and dementia. The disease affects around 1 in 10,000 men and women of all races in their forties. Everyone possesses the gene that causes Huntington's disease; however, only those who inherit the gene's expansion will develop the disease and pass it on to their descendants. Because Huntington disease is inherited by an autosomal dominant gene, any child of a parent with the condition has a 50% chance of acquiring it. "No treatments can alter the course of Huntington's disease" As a result, the emphasis is on improving quality of life with accessible medicines and supportive therapy [1].

### 1.1 Aim

The aim of the study reveals the patients history, prognosis, behavioral, mental, and motor abnormalities are all factors in its clinical and etiopathogenic presentation.

### 1.2 Presentation of Case

Following an inquiry, a 57 -year-old man was taken to Acharya Vinoba Bhave Rural Hospital Wardha on the 15th of March, with the main complaint of difficulties handling and grasping things at his employment as a teacher. Apathy and emotional outbursts were also reported as cognitive consequences.

### 1.3 Present Medical History

Following genetic testing, CAG repeat expansion in Huntington gene (HTT) also called IT-15 gene is determined by two separate PCR assays, – this assays determines the size of the triplet repeat. The patient had 15/41 CAG repeat determines the expanded allele shown in TP-PCR and denotes the full penetrance and diagnosis has been confirmed of Huntington's disease, where, A 57 -year-old female was brought to Acharya Vinoba Bhave Rural Hospital Wardha on the 15th of March, with the major complaint of difficulties handling and holding items at his employment as a teacher as trembling movement all over the body and imbalancing while walking. Apathy and emotional outbursts were also reported as

cognitive consequences. On admission, the patient talked with increased volume and pace.

### 1.4 Past Medical History

Two years ago, the patient notified his family doctor that he was feeling abnormally melancholy on most days and that he was no longer enjoying any happiness in her life. Family history: The patient was alright in the past 50 years before. Six months before the patient was diagnosed with Huntington's disease, her mother had a TIA (Transient Ischemic Attack). She has 2 son and 2 daughters which they are all married and doctor advised them to do the genetic test regarding Huntington's disease which 39 year old daughter has positive for the test. Doctor has advised the treatment on discharge Tab Revocon 25 mg 2 times a day and Tab- Sizodon 0.5 mg once a day and follow-up after 7 days.

## 2. ETIOLOGY

Huntington's disease is caused by an autosomal dominantly inherited CAG trinucleotide repeat expansion in the Huntington (HTT) gene on chromosome [2]. Expansion of an unstable polymorphic trinucleotide (CAG) repeat in axon 1 of the HTT gene or the IT 15 gene, which results in an expanded polyglutamine tract in the protein, is thought to induce a dominant gain of function, resulting in neuronal malfunction and neurodegeneration [3]. Symptoms are probable if it occurs 40 times or more. This alteration leads in a more lethal version of the huntingtin protein. It destroys particular brain cells as it accumulates in the brain.It weakens and finally destroys their function. Scientists also not sure how this happens [4].

### 2.1 Clinical Diagnosis

The clinical presentation of symptoms, a positive family history, and the presence of the genetic marker CAG repeats in HTT are used to make the diagnosis. CAG-Expanded allele and full penetrance. Before motor complaints emerge, CT scan or MRIs scans indicate symmetrical striatal atrophy.

### 2.2 Pathological Discussion

Huntington's disease is a chronic, progressive hereditary disorder defined by the early death of

cells in the striatum of the basal ganglia, a deep within the brain area crucial in movement control [5]. Cells are also lost in the cortex, which is associated with thinking, memory, perception, judgement, and action, as well as in the cerebellum, which is located at the top of the brain. The CAG repeat expansion is unstable and can lengthen during transmission from parents to offspring. Thus age can decrease the from one generation to the next this called as anticipation [6].

### 2.3 Physical Examination

The patient's hands and fingers exhibit uncontrolled, uncontrollable writhing and flexion/extension movements on both sides. He also has mild twitching of his ankles on both sides. With a little thoracic kyphosis, the posture is upright and symmetrical. Gait is characterized by bilateral symmetrical limb movement; cadence and speed were usual. Medical monitoring is required. Because there is no therapy that can stop or reverse the underlying condition, the emphasis is on enhancing quality of life via the use of affordable medications and supportive care. Tetrabenazine is the only medicine licenced by the Food and Drug Administration to treat the symptom of chorea (Xenazine Benzodiazepines and neuroleptic medications have also been found to help in chorea management [7]. Motor signals must be examined and analyzed on a continuous basis in order to achieve appropriate therapeutic medication levels. Akathisia (physical restlessness) in an overmedicated patient is dangerous since it might be mistaken for the illness's restless fretting and therefore

disregarded. Antiparkinson medicines, such as levodopa, may provide some temporary relief in individuals who present with stiffness It has been suggested that tricyclic antidepressants and selective serotonin reuptake inhibitors be used [8].

### 2.4 Nursing Management

Patients with Huntington's disease may be admitted to the hospital for problems linked to the disease and developed a plan of care according to the needs of the patient and implement and evaluate the care.

1. **Safety:** Avoid injuries and potential skin breakdown. Clean the skin well, and stimulate ambulation to assist maintain muscle tone. While feeding, keep the patient as upright as feasible. While feeding, gently support the patient's head with one hand.
2. Medication regimen activities and possible adverse effects must be taught, evaluated, and modified to achieve the desired patient response.
3. **Family education:** The nurse must educate and assist the patient and family as they adjust to the necessary lifestyle adjustments, such as the illness worsens, patients and their families may require home care, case management, counselling, or even legal action to meet the patient's needs. In patients and their families, nurses should check for coping issues, neglect, or abuse. Educate them regarding moderate exercise on a regular basis can help to decrease tightness and spasms [9].



Scanned with CamScanner

Fig. 1. CT scan or MRI scans indicate symmetrical striatal atrophy

### 3. DISCUSSION

A 57-year-old man has been diagnosed with early-stage Huntington's disease. During the first visit, his relatives reported behavioral signs such as indifference as well as chorea in the distal upper extremities. Treatment designed to improve patient stated functional activities such as keyboard typing or blackboard writing (e.g., handwriting task) [10]. Because Huntington's disease is a progressive degenerative condition, therapies that may be useful to the patient in the future were also included in the interventions (e.g., aerobic exercise, respiratory muscle) [11]. The following therapies were conducted at the patient's initial visit: respiratory training, strength training, positioning and balance training, teaching on energy conservation during movement, and training on functional reach and grasp exercises. Balance and postural muscle training were initiated for the same reason: to improve functional mobility and minimize fatigue risks in anticipation of future respiratory muscle degradation. [12]. According to physicians, physiotherapy intervention in the early to middle stages of Huntington's disease can improve motor and functional performance and may even stop disease progression. As a consequence, implementing these treatments early in the disease will almost always assist the patient [13,14-23].

### 4. CONCLUSION

Huntington's disease is a chronic, hereditary nervous system ailment characterized by increasing uncontrollable choreiform movement. The disease affects around 1 in 10,000 men and women of all races in their forties. In this disease physiotherapy intervention may improve the patients self-efficacy, mindfulness and proper gait and balancing. The progressive nature of Huntington's disease demands multidisciplinary treatment and ongoing participation of care in order to provide the best possible assistance to the patient and their family.

### CONSENT AND ETHICAL APPROVAL

The writers have acquired and saved patients' permission and ethical approval in accordance with international standards or university standard guidelines.

### COMPETING INTERESTS

Authors have declared that no competing interests exist.

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