

(CASE REPORT)



Huntington's disease: A comprehensive case report

MALLAMPATI VARSHINI *

Department of Pharmacy Practice, Chalapathi institute of pharmaceutical sciences, Guntur, Andhra Pradesh, India.

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Abstract

Chorea form movements, neuropsychiatric symptoms, and cognitive impairment are hallmarks of Huntington's disease, an inherited progressive neurodegenerative illness that results in severe functional impairment. Between the ages of 30 and 40, it typically shows up. The genetic components of the disease are inherited, meaning that the afflicted individual receives the gene from a parent with the same genetic makeup. It is inherited in an autosomal-dominant manner. It is brought on by increased cytosine-adenine-guanine (CAG) tri-nucleotide repeat in Huntington's (HTT) gene on chromosome 4p. A patient with a strong family history of Huntington's disease is described, associated with their clinical and genetic features.

Keywords: Huntington's disease; Supportive therapy; Polymorphic tri nucleotide; Caudate head atrophy

1. Introduction

Huntington's disease (HD) is a rare hereditary neurodegenerative disorder characterized by chorea, cognitive decline, and psychiatric disturbances ⁽¹⁾. Four to ten cases per 100,000 people are thought to be prevalent in the Caucasian population. Huntington's disease is inherited by an autosomal dominant gene; any child of a parent with the condition has a 50% chance of acquiring it. Huntington's disease is caused by an autosomal dominantly inherited CAG trinucleotide repeat expansion in the Huntington (HTT) gene on a chromosome. 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. Symptoms are probable if it occurs 40 times or more. This alteration leads to a more lethal version of the Huntington protein. It destroys particular brain cells as it accumulates in the brain. In 5–10% of all HD cases, the disease occurs before the age of 21; it is then called juvenile Huntington's disease (5). Huntington's illness cannot be rectified with any treatment. As a result, the emphasis is on improving quality of life with accessible medicines and supportive therapy (2).

2. Case description

This case involves a 22-year-old female patient who was diagnosed with Huntington's disease and admitted to the neurology department; nevertheless, she has not been taking her medicine as prescribed. According to medication compliance, all four limbs' symptoms have returned for the past four years. Caudate head atrophy, which impacts the basal ganglion, is seen on an MRI. Her mother has a positive family history of the illness. Testing for genetics validates the diagnosis. The laboratory's parameters were normal. With the combination of all these data, Huntington's illness is the definitive diagnosis.

* Corresponding author: MALLAMPATI VARSHINI

3. Conclusion

An inherited, long-term neural system disorder called Huntington's disease is marked by an increase in uncontrollable chorea form activity. Due to the degenerative nature of Huntington's disease, patients and their families require continuing care and multidisciplinary treatment to get the best support possible.

Compliance with ethical standards

Statement of informed consent

Informed consent was obtained from the individual participants included in the study.

References

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