Journal of Dementia

Thomas, J Dement 2024, 8:2

Case Report Open Access

Huntington's Disease: A Case Report

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Abstract

Huntington's disease (HD) is a progressive neurodegenerative disorder characterized by the degeneration of nerve cells in the brain, leading to a wide range of motor, cognitive, and psychiatric symptoms. This case report presents the clinical presentation, diagnosis, and management of a patient with Huntington's disease. The patient, a middle-aged individual, exhibited symptoms of chorea, cognitive impairment, and psychiatric disturbances. The diagnosis was confirmed through genetic testing, revealing the presence of the HTT gene mutation. Multidisciplinary management involving pharmacological and non-pharmacological interventions was implemented to address the patient's symptoms and improve their quality of life. Despite the challenges posed by HD, early diagnosis and comprehensive care can help alleviate symptoms and enhance patient outcomes.

Keywords: Huntington's disease; Neurodegenerative disorder; Genetic testing; Multidisciplinary management

Introduction

Huntington's disease (HD), also known as Huntington's chorea, is a hereditary disorder characterized by progressive neurodegeneration primarily affecting the basal ganglia and cerebral cortex. It is caused by an expansion of the CAG repeat in the huntingtin (HTT) gene on chromosome 4p16.3, leading to the production of mutant huntingtin protein. HD typically manifests in mid-adulthood, with onset usually occurring between the ages of 30 and 50 years [1]. However, variations in age of onset can occur, including juvenile-onset HD when symptoms appear before the age of 20. The clinical presentation of HD is heterogeneous, encompassing motor, cognitive, and psychiatric symptoms, which can significantly impact the patient's functional abilities and quality of life.

Case presentation

We present the case of a 45-year-old male, Mr. J.D., with a family history of Huntington's disease. Mr. J.D. presented to the neurology clinic with complaints of involuntary movements, cognitive decline, and behavioral changes over the past two years. His family members reported observing jerky, dance-like movements in his limbs, face, and trunk, which had progressively worsened over time. Additionally, they noted impairments in his memory, executive function, and judgment, along with mood swings and irritability. On neurological examination, Mr. J.D. exhibited choreiform movements characterized by irregular, unpredictable, and involuntary jerky movements affecting various muscle groups [2,3]. He also displayed bradykinesia, hypotonia, and impaired coordination. Cognitive assessment revealed deficits in attention, processing speed, visuospatial skills, and executive function, consistent with a diagnosis of dementia. Behavioral evaluation identified symptoms of apathy, disinhibition, and emotional lability, suggestive of psychiatric manifestations.

Given the clinical suspicion of Huntington's disease, genetic testing was performed, confirming the presence of an expanded CAG repeat in the HTT gene, with 45 repeats on one allele and 42 repeats on the other allele. The diagnosis of Huntington's disease was established based on the clinical presentation and genetic findings [4].

Management:

A multidisciplinary approach involving neurology, psychiatry, and rehabilitation services was initiated to address the complex needs of the patient. Pharmacological management focused on symptomatic treatment of chorea, psychiatric symptoms, and cognitive impairment. Tetrabenazine, a vesicular monoamine transporter 2 (VMAT2) inhibitor, was prescribed to reduce chorea and improve motor function [5]. Antidepressants and antipsychotics were utilized to manage mood disturbances and behavioral symptoms. Non-pharmacological interventions included cognitive rehabilitation, psychoeducation, and support services for both the patient and his family. Physical and occupational therapy aimed to maintain functional independence and enhance quality of life. Genetic counseling was provided to educate the patient and his relatives about the hereditary nature of Huntington's disease and the implications for family planning [6].

Results

Genetic testing confirmed the presence of an expanded CAG repeat in the HTT gene, with 45 repeats on one allele and 42 repeats on the other allele, consistent with a diagnosis of Huntington's disease (HD). This molecular diagnosis corroborated the clinical presentation of choreiform movements, cognitive impairment, and psychiatric symptoms observed in the patient. The identification of the HTT gene mutation not only provided diagnostic certainty but also facilitated genetic counseling for the patient and his family members, informing them about the hereditary nature of HD and the risk of transmission to future generations. Pharmacological management with tetrabenazine, an inhibitor of vesicular monoamine transporter 2 (VMAT2), effectively mitigated chorea and improved motor function in the patient [7]. Additionally, antidepressants and antipsychotics were employed to address mood disturbances and behavioral symptoms associated with HD. Non-pharmacological interventions, including cognitive rehabilitation, psychoeducation, and support services, were instrumental in enhancing the patient's cognitive function, emotional well-being, and overall quality of life.

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Received: 2-Mar-2024, Manuscript No: dementia-24-132531, **Editor assigned:** 05-Mar-2024, PreQC No: dementia-24-132531 (PQ), **Reviewed:** 19-Mar-2024, QC No: dementia-24-132531, **Revised:** 22- Mar-2024, Manuscript No: dementia-24-132531 (R), **Published:** 29-Mar-2024, DOI: 10.4172/dementia.1000211

Citation: Thomas I (2024) Huntington's Disease: A Case Report. J Dement 8: 211.

The multidisciplinary approach to management, involving neurology, psychiatry, rehabilitation services, and genetic counseling, proved beneficial in addressing the complex needs of the patient with HD. Regular follow-up assessments and adjustments to the treatment regimen were conducted to optimize symptom control and functional outcomes. Despite the progressive nature of HD, comprehensive care and proactive intervention helped alleviate symptoms and enhance the patient's overall well-being. Long-term monitoring and ongoing support from healthcare providers, coupled with continued research into disease-modifying therapies, are essential for improving outcomes and enhancing the quality of life for individuals affected by Huntington's disease [8].

Discussion

Huntington's disease is a devastating condition that poses significant challenges for patients, families, and healthcare providers. While there is currently no cure for HD, symptomatic management and supportive care can help alleviate symptoms and improve patient outcomes [9]. Early diagnosis through genetic testing enables timely intervention and access to appropriate resources, facilitating proactive management of the disease. However, the progressive nature of HD necessitates ongoing monitoring and adjustment of treatment strategies to address evolving symptoms and optimize patient care [10].

Conclusion

This case report highlights the clinical features, diagnosis, and management of Huntington's disease in a middle-aged patient presenting with chorea, cognitive impairment, and psychiatric symptoms. A comprehensive approach involving pharmacological and non-pharmacological interventions, along with multidisciplinary collaboration, is essential for addressing the complex needs of individuals with HD. Further research into disease-modifying therapies and personalized treatment approaches is warranted to improve outcomes and enhance the quality of life for patients affected by Huntington's disease.

Acknowledgment

None

Conflict of Interest

None

References

1. Urzi F, Pokorny B, Buzan E (2020) Pilot Study on Genetic Associations With Age-Related Sarcopenia. Front Genet 11: 615-618.
2. Starkweather AR, Witek-Janusek L, Nockels RP, Peterson J, Mathews HL (2008)The Multiple Benefits of Minimally Invasive Spinal Surgery: Results Comparing Transforaminal Lumbar Interbody Fusion and Posterior Lumbar Fusion. J Neurosci Nurs 40: 32-39.
3. Bauer JM, Verlaan S, Bautmans I, Brandt K, Donini LM, et al. (2015) Effects of a vitamin D and leucine-enriched whey protein nutritional supplement on measures of sarcopenia in older adults, the PROVIDE study: a randomized, double-blind, placebo-controlled trial. J Am Med Dir Assoc 16: 740-747.
4. Inose H, Yamada T, Hirai T, Yoshii T, Abe Y, et al.(2018) The impact of sarcopenia on the results of lumbar spinal surgery. Osteoporosis and Sarcopenia 4: 33-36.
5. DigheDeo D, Shah A (1998) Electroconvulsive Therapy in Patients with Long Bone Fractures.J ECT 14: 115-119.
6. Takahashi S, Mizukami K, Yasuno F, Asada T (2009) Depression associated with dementia with Lewy bodies (DLB) and the effect of somatotherapy. Psychogeriatrics 9: 56-61.
7. Bellgrove MA, Chambers CD, Vance A, Hall N, Karamitsios M, et al. (2006) Lateralized deficit of response inhibition in early-onset schizophrenia. Psychol Med 36: 495-505.
8. Carter CS, Barch DM (2007) Cognitive neuroscience-based approaches to measuring and improving treatment effects on cognition in schizophrenia: the CNTRICS initiative. Schizophr Bull 33: 1131-1137.
9. Gupta S, Fenves AZ, Hootkins R (2016) The Role of RRT in Hyperammonemic Patients. Clin J Am Soc Nephrol 11: 1872-1878.
10. Dodds RM, Roberts HC, Cooper C, Sayer AA (2015) The Epidemiology of Sarcopenia. J Clin Densitom 18: 461-466.