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PHYSIOTHERAPEUTIC INTERVENTION IN HUNTINGTON'S DISEASE – CASE STUDY

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ABSTRACT. *Introduction:* Huntington's disease is a hereditary neurodegenerative disorder characterized by motor, cognitive, and behavioral symptoms, with onset between 30 and 50 years of age. *Objectives:* The aim of this study was to evaluate the impact of physiotherapy on mobility, motor control, and balance, aiming to improve the quality of life of the patient in daily activities. *Material and Methods:* The study was conducted on a 47-year-old patient diagnosed with Huntington's disease at age 39. Over 10 weeks, with 3 weekly physiotherapy sessions, four assessment stages were performed using the following scales: Mini-Mental State Examination for cognition, Berg and Tinetti Scale for balance, Fahn-Marsden Scale for dystonia, and Activities of Daily Living for daily activities. *Discussions:* The results indicated significant improvements in balance and motor control, as well as a slight improvement in dystonic movements and independence in daily activities, especially in personal hygiene. *Conclusions*: The study concludes that physiotherapy interventions can slow the progression of symptoms and contribute significantly to improving the quality of life of patients with Huntington's disease, highlighting the importance of this therapy.

Keywords: Huntington's disease; physical therapy; involuntary movements; quality of life.

INTRODUCTION

Huntington's disease, first described by Waters in 1842 and detailed by George Huntington in 1872, is a neurodegenerative disorder with onset in middle age, characterized by involuntary movements, cognitive and behavioral disorders, and dementia. In 1993, the discovery of the HTT gene on chromosome

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4 revolutionized the understanding and treatment of the disease, becoming a model for medical research. The disease is caused by the expansion of the CAG trinucleotide in the HTT gene, which produces the mutant huntingtin protein, responsible for neuronal toxicity. It is an autosomal dominant condition, with a 50% probability of being transmitted to offspring (Bates, 2015; Roos, 2010).

Mutant huntingtin protein affects neuronal structure and function through abnormal aggregation, mitochondrial dysfunction, disruption of axonal transport, and alterations in gene expression. The secretion of neurotransmitters such as dopamine, glutamate, and GABA is imbalanced, leading to damage to the basal ganglia and cortex, which are involved in movement control and cognitive functions. The first molecular signs are detectable 15 years before the onset of clinical symptoms (Bates, 2015; Irfan, 2022).

Motor symptoms include chorea, bradykinesia, rigidity, and dystonia. Cognitive disorders are manifested by slowed thinking, memory impairment, and difficulty in planning. Psychiatrically, patients may present with depression, anxiety, apathy, and behavioral disorders, often preceding motor symptoms. The disease progresses progressively, leading to total dependence and severe disability in advanced stages (Caron et al, 1998; Roos, 2010).

Diagnosis is based on: medical and neurological history (analysis of reflexes, coordination and mental status), imaging (MRI/CT reveals striatal and cortical atrophy), and genetic testing (identifies CAG extensions in the HTT gene, considered the defining criterion) (Bates, 2015; National Institute of Neurogical Disorders and Stroke, 2025).

Direct genetic testing is the reference method, complemented by imaging investigations and analyses to exclude other conditions. The symptomatic triad (motor, cognitive and neuropsychiatric disorders) defines the disease, being supported by visual and olfactory abnormalities (National Institute of Neurogical Disorders and Stroke, 2025).

Huntington's disease is a complex condition with multisystemic impact. Current research allows for early diagnosis and opens up therapeutic perspectives, especially in preclinical stages (Bates, 2015).

Huntington's disease is progressive, with a median survival of 15–25 years after onset. Death is frequently due to pneumonia, cardiovascular complications, and conditions such as choking, fractures, and suicide. Choreic movements increase the risk of head trauma (Caron et al, 1998, Liou, 2010)

Although there is no cure, patients are treated with various therapeutic interventions, taking into account their symptoms. Thus, interventions can be made with: **physical therapy** - for maintains mobility, preventing contractures, and improving balance through adapted exercises in order to reduce the risk of falls, and correcting gait and posture (Fritz et all, 2017, Quinn, L., & Rao, A. 2002); **occupational therapy** – for promoting the independence through environmental

adaptation and the use of assistive devices (Bilney et al, 2003; Simon-Vicente et al, 2023); **speech therapy** - addresses dysphagia and speech disorders through muscle exercises and nutritional guidance (Liou, 2010); p**sychotherapy** - focuses on managing depression, anxiety, apathy, and other cognitive and behavioral problems and cognitive behavioral therapy is effective in obsessive-compulsive disorders (Zanotti et al, 2020); **drug treatments** – motor and cognitive symptoms are managed with medications, but their choice must balance efficacy and the risks of adverse effects (Liou, 2010).

Patients affected by this rare degenerative disease often face considerable difficulties in obtaining adequate care and effective treatments. This condition presents a distinct challenge, as its diagnosis and management often require specialized expertise and specific resources.

In this paper, we aimed to follow the evolution of a subject diagnosed with Huntington's disease following the implementation of a physiotherapy program. The aim of the intervention was to monitor changes in mobility, motor control and balance following the physiotherapy program and the effects on the patient's quality of life in daily activities. Physiotherapy could contribute to reducing choreic movements, improving the patient's balance, mobility and functional autonomy. It also explores the potential of physiotherapy to slow the progression of the disease, stimulating motor and neurological functions and facilitating the patient's adaptation to the changes imposed by the condition.

MATERIAL AND METHODS

Participants

The case study is conducted on a female subject, aged 47, diagnosed with Huntington's Disease at the age of 39. 10 years ago, she presented to the county hospital in her locality in the emergency department with psychomotor agitation, behavioral disorders, insomnia and prevalent ideas. She was discharged with the diagnosis of Organic Delusional Disorder and Mental Retardation with Behavioral Disorders. After 2 years of constant hospitalizations caused by behavioral disorders and episodes of depression resulting in aggressive behavior manifested both towards those around her and towards herself, she was diagnosed with Huntington's Chorea.

In 2022, she was institutionalized in a center for the care of people with disabilities. Over time, she did not benefit from physiotherapy sessions or other forms of therapy, being prescribed exclusively drug treatment.

The therapeutic intervention was carried out with the written consent of the relatives and with the written agreement of the management of the institution where the subject is institutionalized.

Materials

The study was conducted over a period of 18 weeks, within the institution where the patient is hospitalized. After an initial assessment, therapeutic interventions were postponed for 8 weeks, providing the opportunity to observe the natural evolution of the disease. Subsequently, a personalized physiotherapy program was implemented for 10 weeks, with assessments every 5 weeks. Various assessment tools, such as Mini-Mental State Examination (MMSE), ADL Scale, Berg Scale, Tinetti Scale and Fahn-Marsden Scale, were used to monitor progress.

The physiotherapy program included prehension exercises, balance, gait and supine exercises, based on the Frenkel program. The main objectives were to improve balance, coordination, clinical manifestations, quality of life and promote independence in daily activities.

RESULTS

The 18-week study included four assessments: baseline, after 8 weeks without intervention, intermediate after 5 weeks of physiotherapy treatment and final after another 5 weeks of therapy. At the beginning, the patient had moderate balance, a medium risk of falling, a degree of assisted independence and relatively good cognitive function, but with difficulties in calculation, writing and drawing. The degree of dystonia was moderate, allowing her to carry out her daily activities. Therapeutic intervention did not begin immediately after the initial assessment due to administrative delays in obtaining the center's approval to carry out the therapy and severe depressive episodes, triggered by a family event, which affected the patient's emotional state. Under these conditions, a second assessment was carried out after 8 weeks without therapeutic intervention, which revealed that the patient's condition had worsened, with regressions in balance, memory and spatial-temporal orientation, and a high risk of falling. After 5 weeks of treatment, improvements were observed in balance, gait, mental status, and dystonia. The final assessment confirmed the progress, with significant improvement from baseline.

Berg Scale

The Berg Scale was used to assess the balance disorders of the patient in the study. The assessment was carried out in four stages, the values of each assessment are presented in the following graph (figure 1).

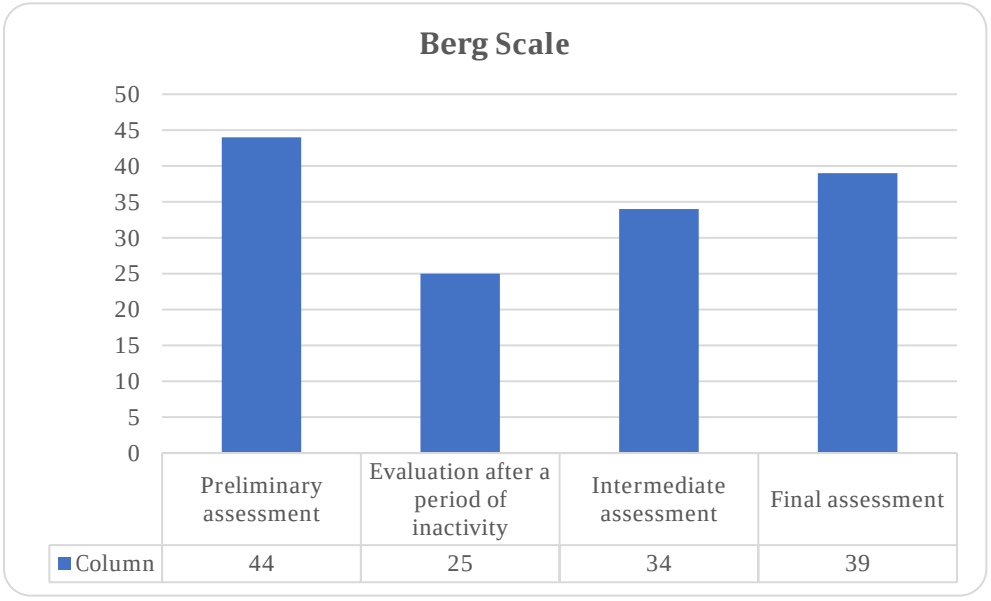

Fig. 1. Berg Scale – graphic representation
(0-20 – severely impaired balance; 21-40 – moderately impaired balance;
41-56 – almost normal balance)

The diagram highlights a severe regression of balance after the period without physiotherapy, followed by a significant improvement after 5 weeks of treatment, with continued improvements in the following period.

Tinetti Scale

To illustrate the patient's progress during the therapeutic intervention, we used the Tinetti scale, a validated and recognized tool for assessing balance and gait. The graph below (figure 2) presents the values obtained from periodic assessments performed throughout the study.

At baseline, the patient had a Tinetti score of 19, indicating moderate balance and stability, with moderate risk of falling. After 8 weeks without therapy, the score dropped drastically to 8, reflecting significant deterioration. The physiotherapy intervention led to an increase in the score to 20 after 5 weeks of treatment and to 21 at the end of 10 weeks, highlighting an almost complete restoration of balance and stability.

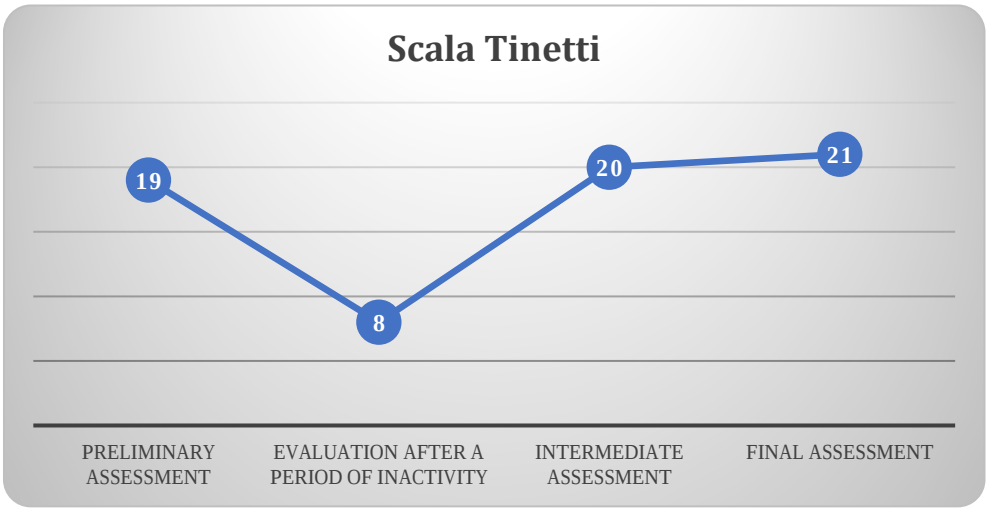

Fig. 2. Tinetti Scale – graphic representation
(< 19 – high risk of falling; 19-20 – moderate risk of falling; > 25 – low risk of falling)

MMSE (Mini Mental State Examination)

The patient's cognitive function was assessed with the MMSE, and the graph (figure 3) illustrates the evolution of the scores, highlighting the changes during the therapeutic intervention.

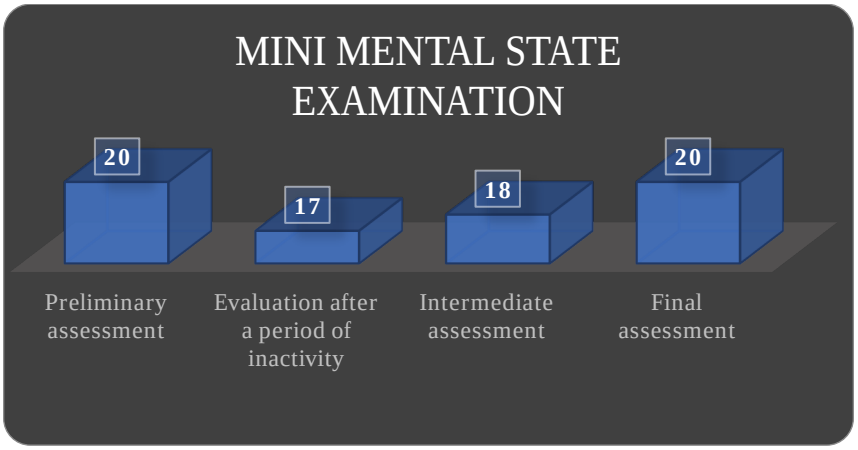

Fig. 3. Mini Mental State Examination – graphic representation
(> 21 mild cognitive dysfunctions; 0-20 moderate cognitive dysfunctions;
9 severe cognitive dysfunctions)

At baseline, the patient had an MMSE score of 20, indicating mild cognitive impairment. The score had decreased to 17 before therapy, reflecting deterioration with disorientation and difficulty following commands. After physiotherapy, the score increased to 18 and eventually returned to 20, indicating restoration of baseline, with improvements in orientation, information retrieval, and language. Attention, calculation, and writing remained impaired.

Activities of Daily Living (ADL) Scale

The data collected during the assessment of activities of daily living are illustrated in figure 4.

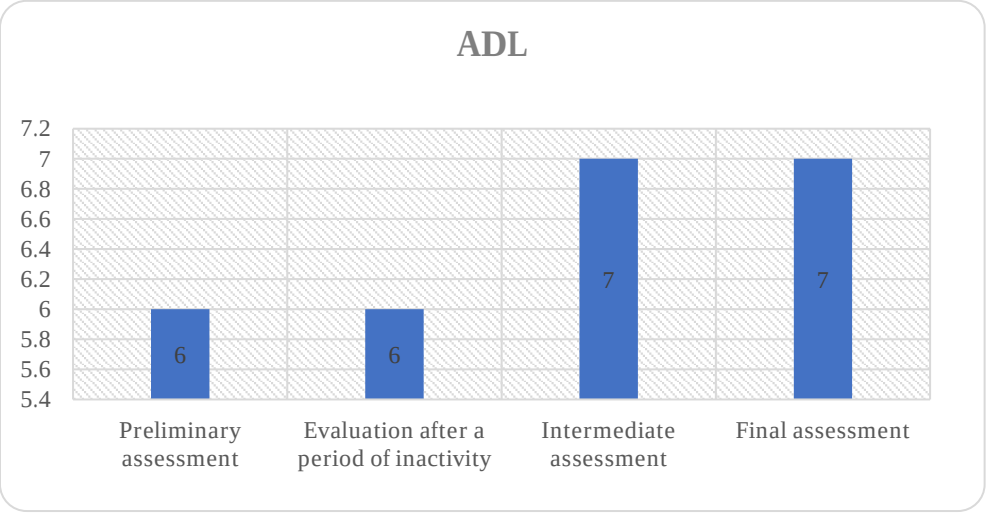

Fig. 4 ADL scale – graphic representation
(10 – autonomy; 8-10 – quasi-dependent; 3-8 – assisted independence; 0-3 – total dependence)

At the initial assessment, the patient obtained a score of 6, indicating assisted independence. The score remained unchanged before therapy. After the physiotherapy intervention, the score increased to 7, with improvements in personal hygiene, where she went from total dependence to partial help. The score was maintained at 7 at the final assessment, highlighting a modest but significant progress for quality of life.

Fahn-Marsden Scale

To assess the severity and frequency of involuntary movements in the patient in the study, we used the Fahn-Marsden scale, a specialized instrument for measuring the degree of dystonia. The values obtained from the assessments with the Fahn-Marsden scale are presented in figure 5.

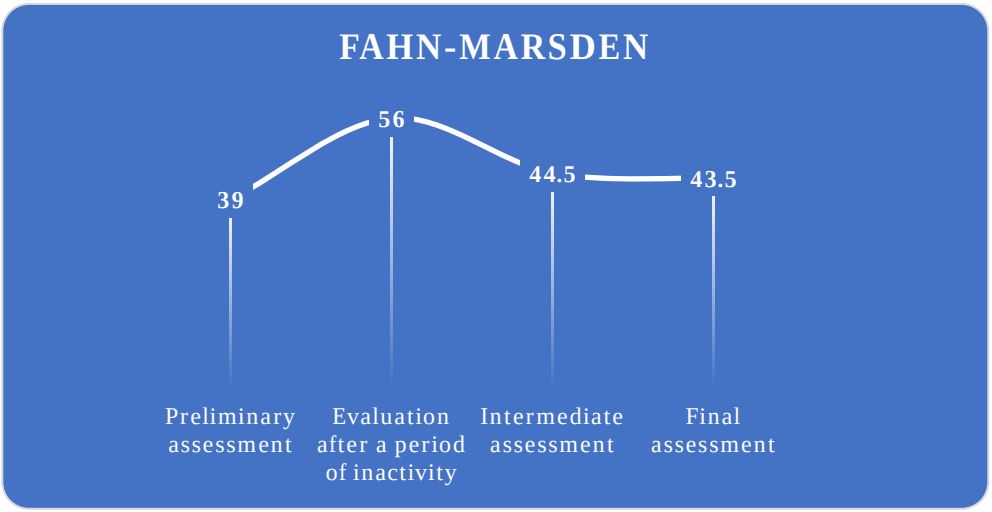

Fig. 5 Fahn-Marsden scale – graphic representation

At the initial assessment, the score of 39 indicated a moderate degree of dystonia, without major impact on daily activities. The score increased to 56 before therapy, reflecting a significant worsening. After physiotherapy, at the intermediate assessment the score decreased to 44.5, and at the final assessment to 43.5, indicating a progressive improvement in movement control and frequency of dystonic episodes.

DISCUSSION

Patients with Huntington's Disease suffer from involuntary movements (chorea), coordination difficulties, speech and swallowing problems, cognitive decline and significant personality changes; symptoms that progressively worsen, affecting daily activities and independence. The present study demonstrates the positive effects of physiotherapy on the patient studied.

At the initial assessment, the patient presented good balance (Tinetti score 19), assisted independence (ADL score 6), moderate cognitive function (MMSE 20) and moderate dystonia (Fahn-Marsden 39). After 8 weeks without intervention, the condition deteriorated: Tinetti score 8, MMSE 17, Fahn-Marsden 56, while ADL remained constant at 6, demonstrating a deterioration in balance, gait, cognitive function and a worsening of dystonic manifestations.

The physiotherapy intervention had a positive impact on the patient's condition. After the first 5 weeks of treatment, interim assessments showed significant improvements: the Tinetti score increased to 20, the MMSE to 18, and the Fahn-Marsden decreased to 44.5. These changes suggest an improvement in balance, gait, cognitive function, and dystonic manifestations. The ADL scale showed a slight improvement, with the score increasing to 7, reflecting a greater capacity for self-care, especially in the personal hygiene category. Progress continued in the following weeks, with results highlighting improvements in balance, motor, and cognitive function, with a positive impact on quality of life.

These findings are supported by the literature. Quinn et al. (2014) demonstrated that regular physiotherapy intervention reduces the risk of falls and improves motor and cognitive function. Chang et al. (2012) highlighted the role of structured physical exercise in preventing cognitive decline in elderly patients with neurological disorders. Piira et al. (2014) also showed that long-term multidisciplinary programs can improve gait, balance, and psychological well-being in patients with Huntington's disease, although the effects on cognitive function and ADL are minor.

Limitations of the study include the institutionalization of the patient, which may influence the results, and the fact that the research focused on a single case, restricting the generalizability of the conclusions. Also, the short period of therapy limits the observation of long-term effects. Future studies will explore multidisciplinary interventions to obtain more relevant results regarding the comprehensive management of this condition.

CONCLUSIONS

Huntington's disease is a progressive neurodegenerative disorder that affects motor, cognitive and mental functions, having a significant impact on the autonomy and quality of life of patients, but also on caregivers. Physiotherapy plays an important role in the management of this disease, providing benefits such as improved balance, coordination and reduced risk of falls, thus maintaining a degree of independence in daily activities.

The effectiveness of physiotherapy intervention was demonstrated by significant improvements in balance and gait (measured by the Berg and Tinetti scales), reduction of dystonia (Fahn-Marsden Scale) and stabilization of cognitive functions. The intervention also led to an improvement in orientation, language and the ability to reproduce information, contributing to better autonomy and quality of life.

In the absence of physiotherapy, rapid progression of the disease was observed, manifested by the degradation of motor and cognitive functions, highlighting the need for constant treatment. Through personalized exercises, physical therapy slows functional decline, stimulates neural connections, and provides psychosocial support, reducing anxiety and isolation.

Physical therapy is essential in the management of Huntington's disease, bringing significant physical, cognitive, and emotional benefits, supporting families and caregivers in managing daily tasks.

AUTHOR CONTRIBUTIONS

Conceptualization and methodology RMG and AMV; planning the therapeutic intervention AMV and CMP; investigation RMG; implementing the therapeutic intervention RMG; writing and data analysis RMG, AMV and CMP. All authors contributed equally to this article.
All authors have read and agreed to the published version of the manuscript.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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