PATIENT CARE FOR INDIVIDUALS WITH HUNTINGTON'S DISEASE

Leonarda-Ana Poljak[1] & Sabina Bis[2]

[1] Karlovac Pharmacy, 47 000 Karlovac-Dubovac, Croatia

[2] Bjelovar University of Applied Sciences, 43 500 Daruvar, Croatia

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ABSTRACT

Introduction: Huntington's disease is a progressive autosomal dominant neurodegenerative disorder characterized by a triad of clinical features affecting an individual's cognitive, physical, and psychological functions. Symptoms typically manifest in middle age, between 30 and 40 years, leading to death within one to two decades after the onset of neurological deterioration. This incurable disease renders patients entirely dependent on care, significantly impacting family members, friends, and close associates. Patient care focuses on alleviating symptoms, maintaining functional ability, and preserving quality of life. It is provided by a multidisciplinary healthcare team that includes pharmacological and nonpharmacological interventions.

Objective: This paper aims to conduct a literature review and present a comprehensive overview of nursing care processes for patients with Huntington's disease.

Conclusion: Guidelines for caring for patients with Huntington's disease provide a foundation for a multidisciplinary approach and enhance patient quality of life, emphasizing comfort, safety, and dignity. As key team members, involving specialized nurses contributes to reducing hospitalization durations, optimizing care levels, and improving the quality of life for patients with Huntington's disease.

Keywords: patient, Huntington's disease, patient care.

Correspondence author: Sabina Bis, sbis@vub.hr

INTRODUCTION

Huntington's disease (HD) is defined as a chronic, neurodegenerative, hereditary disorder causing the degeneration of nerve cells within the brain. The middle age range of 30–40 years is the most common period for disease onset, irrespective of gender or race. However, juvenile-onset HD, appearing before age 21, is also possible. The disease affects movement, behavior, personality, memory, cognition, learning, and comprehension, with chorea being the most common symptom. The cause of HD is a gene mutation on the short arm of chromosome 4, and the diagnosis can be confirmed through genetic testing (1).

Since the disease is incurable, treatment is symptomatic, involving both pharmacological and non-pharmacological methods. As a member of a multidisciplinary team, nurses perform numerous interventions focused on providing palliative care. Given the significant impact of HD on the lives of patients and their families, the nurse's role is to offer support, uphold patient dignity and autonomy, and ensure an optimal quality of life (2).

This paper aims to showcase the role of the nurse within the multidisciplinary approach to nursing care for patients with Huntington's disease. Publications in scientific databases like PubMed, Hrčak, Medline, Google Scholar, CROSBI, BMJ Journals, ScienceDirect, Scopus, NIH, and Sci-Hub were reviewed using keywords: Huntington's disease, chorea, movement disorders, and nursing care, in both Croatian and English. Despite the recognized need for specialized nurses to improve the quality of care for HD patients, there is a lack of research papers specifically addressing HD patient care. Two caregiving guides featuring case studies provided the most comprehensive overview of care for HD patients.

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Care Guidelines are joint documents developed by experts to optimize care standards within specific geographic regions. These guidelines provide standards of care and promote uniform quality through broad dissemination of their content (2). Given the considerable variability in HD symptomatology, appropriate patient care requires a multidisciplinary approach involving a team of specialists: a general practitioner, psychiatrist, geneticist, nurse, rehabilitation team, nutritionist, and social work services (2, 3).

Patients with Huntington's disease have specific care needs that require specialized nursing knowledge. HD-specialized nurses reduce hospital admissions (>50%), shorten hospital stays, and improve care quality in hospital and outpatient settings (4). During patient hospitalization, the primary responsibility of the nurse is to assess the patient's needs when establishing a care plan, prioritizing a team approach to meet patient needs (5).

Weight loss in HD patients is common, especially in advanced stages, and may result from metabolic dysfunction, eating and swallowing difficulties, loss of fine motor control, anorexia, or cognitive impairment. The risk of aspiration is notably high for patients on oral diets, particularly in the later stages of dysphagia. Managing food consistency and portion size is an effective strategy for safe swallowing in HD patients. Monitoring patients during feeding, ensuring adequate caloric intake and nutritional supplementation, and consulting with a speech therapist to assess swallowing

abilities and a nutritionist are recommended (6, 7).

Whether a patient consumes food orally or uses alternative feeding methods, such as percutaneous endoscopic gastrostomy, nausea, and vomiting may be problematic. Possible causes of these symptoms, such as infection, medication side effects, constipation, and symptom timing (before/after food or drink consumption), should be explored. Providing ample time and proper positioning during feeding, offering small and frequent meals, and encouraging patients to eat slowly and sip fluids can significantly reduce nausea and vomiting. For patients on tube feeding, checking patient positioning, feeding rate, and food suitability is essential; antiemetic medication should also be considered (8).

Elimination issues, common in neurodegenerative disorders, can significantly reduce the quality of life for HD patients. Overactive bladder, urinary incontinence, and incomplete bladder emptying are prevalent among HD patients, along with diminished anal sphincter tone and voluntary control. These symptoms, reflecting central nervous system degeneration, worsen over time and may result in fecal incontinence or constipation. Depending on disease progression, assistance may be required to maintain hygiene and recognize signs of incontinence. Nursing interventions include timely identification of elimination difficulties and skin integrity maintenance. To ensure comfort and dignity, appropriate products and aids should be used. With chorea, standard pads may be ineffective, and suprapubic catheterization may be the best option. In addition to using pads and bladder catheterization, antimuscarinic medications may be included in the therapy.

Urinary tract infections, unfortunately common in advanced HD patients, result in acute symptom worsening and require immediate treatment. Preventive and treatment interventions include maintaining adequate hydration. Disease progression, increased immobility, effects of therapy, and poor nutrition and hydration status lead to patient constipation, managed through increased fluid and fiber intake and the use of laxatives (5).

Sleep and wakefulness disorders are among the earliest symptoms of many neurodegenerative disorders. Even when patients are in familiar surroundings, they often experience sleep disturbances such as difficulty falling asleep, frequent awakenings, insomnia, and daytime fatigue. If the patient is hospitalized, these issues tend to intensify. Standard strategies to improve sleep, like removing environmental factors that hinder sleep, minimizing noise and light, maintaining routines and daily activities, and keeping a sleep diary, are effective for patients with Huntington’s Disease (HD) and aid them in achieving better rest. Medication and relaxation techniques are also recommended. Improved sleep quality can positively affect cognitive impairment in HD patients, as well as reduce anxiety, depression, and irritability (7).

Progressive changes in mobility are an integral part of the clinical picture in HD patients, leading to decreased independence and an increased risk of falls. Cognitive decline also affects mobility, gait problems, and balance. Safe movement and handling, especially lifting, can be very challenging due to choreatic movements (5). Nurses should implement all interventions aimed at preventing falls and injuries in line with institutional policies, with an emphasis on educating patients about the safe use of

assistive devices and assessing their mobility capabilities. Optimizing function according to disease stage and encouraging participation in self-care activities are key goals of nursing care for HD patients. Stiffness, rigidity, and dystonia can cause significant pain and hinder hygiene maintenance, dressing, and comfortable positioning. Jaw stiffness can further complicate eating, drinking, and oral hygiene. Integrating physical and occupational therapy is imperative in the treatment of HD patients.

In the later stages of Huntington's disease, severe behaviors such as aggression and impulsivity become less problematic as the patient becomes physically less able to act out. However, other behaviors, like resistance, agitation, and screaming, may emerge, and episodes of aggression remain possible, potentially endangering the patient or others. If the patient’s behavior changes suddenly, it is important to check for factors that could worsen symptoms, such as infection or pain, and to rule out head injury. Environmental factors contributing to behavioral changes and issues should be considered. Familiar, calm surroundings, adherence to routines, and allowing adequate time for self-care activities may help with resistance. Unfortunately, the exact cause of the screaming is often not identifiable. In such cases, strategies for managing agitation, providing reassurance and comfort, alleviating possible pain, and reviewing medications that may cause side effects are recommended (7).

In addition to motor and cognitive disorders, Huntington's Disease also causes psychiatric issues. Depression, irritability, and apathy are among the most common psychiatric symptoms. The severity of symptoms varies significantly and can manifest as aggressive outbursts, impulsivity, social withdrawal, and even suicide (4). Patients should be assessed for suicidal thoughts and ideas, and treatment should start with low doses of pharmacological therapy to minimize potential side effects (9). In cases of irritability, staying calm and avoiding conflict is essential, along with trying to identify situations that trigger anger and irritability in the patient. Apathy requires interventions that include encouraging the patient and maintaining routines with calendars and schedules to help with motivation. Psychotherapy should not be dismissed solely because of communication issues and/or cognitive impairment, as it provides many HD patients an opportunity to discuss their feelings, fears, and frustrations, which is highly beneficial (6).

Patients in advanced stages of HD may experience pain for various reasons, though assessing pain can be difficult if the patient is unable to communicate. Using validated pain scales for patients who are non-communicative and/or have dementia can be helpful. Attention should be paid to non-verbal cues and possible sources of pain (e.g., bruises, wounds, fractures). Effective treatment may be achieved with medications such as gabapentin, pregabalin, and amitriptyline. In some cases, opioid analgesics, such as tramadol and fentanyl patches, are necessary. Besides pharmacological methods, non-pharmacological interventions should also be included in the pain management process (5, 6).

As Huntington's disease progresses, the ability to communicate effectively and make decisions declines. Dysarthria develops, and choreatic movements further complicate speech comprehension. Establishing a trusting nurse-patient relationship is crucial for effective

communication (10). Early referral to speech and language therapy enables continuous assessment of communication skills and provides suggestions for alternative communication methods, which may include various symbols, devices, strategies, and techniques to compensate for speech, language, and communication difficulties. Devices used may be low-tech or high-tech, including talking and memory boards, diaries, photo albums, pictorial symbols, and thumb-up/thumb-down responses. Equipment that has proven particularly effective includes devices like the LightWriter and talking mats, which should be adapted to accommodate possible chorea or rigidity (8). During communication, it is essential to provide ample time, remain patient, and minimize background noise, which can be disturbing and distracting to the cognitive process. Attention should be given to non-verbal cues, voice changes, or any sounds the patient can make. Introducing new skills to enable effective communication in the later stages of the disease is sometimes not advisable, as cognitive decline may prevent the patient from developing the skills to use them appropriately. Nurses should understand this complex set of behaviors to adapt their practice accordingly and develop various ways to communicate with the patient.

HD patients have hobbies and interests, and it is essential that they engage in them. Caregivers/family members play a role in facilitating this, and, where possible, the involvement of an activities coordinator can be beneficial. Planning activities based on the patient's interests and considering their shorter attention span and tendency to fatigue are important. Developing and using tools like books and gathering information from family and friends about the patient's hobbies and interests can guide patients toward activities that engage them (7, 8).

Formal involvement of palliative care providers usually begins in the later stages of the disease. Given the nature of disease progression, palliative care should prioritize setting immediate care goals that improve quality of life and prevent caregiver burnout (2). Some HD patients are cared for at home, often by a family member or caregiver. Nurses should assist family members in providing informal care and support, aiming to ensure individualized care for the patient and guiding treatment decisions according to the patient's wishes. For many patients, caregiver loss, inability to live independently, inappropriate behavior, and complex care needs may result in admission to a chronic long-term care facility. For such patients in nursing homes, common issues include smoking, urinary and/or fecal incontinence, pressure ulcers, and skin ulcers.

The comprehensive goals of nursing care are to optimize the comfort, safety, dignity, and autonomy of the patient. These principles should be considered when creating care plans and implementing them (7).

Given the diversity of Huntington's disease symptoms, there is a wide range of nursing diagnoses, the most common of which include (11):

- Risk for injury related to disorientation
- Risk for falls related to cognitive impairment
- Risk for infection related to invasive procedures
- Risk for violent behavior related to delusional thinking
- Risk for aspiration related to dysphagia
- Chronic confusion related to structural/functional changes in brain tissue

- Impaired self-care abilities (feeding, elimination, dressing, bathing) related to cognitive impairment
- Impaired mobility related to musculoskeletal damage, evidenced by uncoordinated movements
- Decreased social interaction

The nurse, as a member of a multidisciplinary team, plays a key role in caring for patients with Huntington's disease (12). The nurse ensures that care plans are developed to maintain the patient's safety and dignity, identify medically significant changes in the patient's condition, communicate these to the physician, and provide ongoing support to the patient and their family throughout the illness. Additionally, the nurse may assume the role of team leader, coordinating with physicians (including family medicine doctors, neurologists, psychiatrists, and palliative care/hospice physicians), dentists, rehabilitation therapists, and support providers (such as psychologists, social workers, and spiritual care providers), as well as other healthcare professionals (dietitians, pharmacists, laboratory technicians, etc.) (7, 13). Each team member has a specific role in the care of Huntington's patients, with the nurse being ideally positioned to determine when their services are needed and prioritize their involvement. Developing treatment guidelines and including patients and their families in high-quality, compassionate care, although challenging, greatly improves the lives of those affected by Huntington's disease. Conclusion Guidelines for caring for HD patients are a crucial tool in ensuring quality and consistent care. Given the diversity of symptoms, a multidisciplinary approach is essential, including doctors from various specialties, nurses, therapists, and other specialists. Specialized nurses play a key role in improving care quality, reducing hospitalizations, and optimizing patients' comfort and autonomy levels. During hospitalization, nurses assess patients' needs and provide appropriate care, particularly in areas such as nutrition, elimination, safety, and communication. Nursing interventions aim to ensure patient comfort, safety, dignity, and autonomy.

CONCLUSION

Guidelines for caring for HD patients are a crucial tool in ensuring quality and consistent care. Given the diversity of symptoms, a multidisciplinary approach is essential, including doctors from various specialties, nurses, therapists, and other specialists. Specialized nurses play a key role in improving care quality, reducing hospitalizations, and optimizing patients' comfort and autonomy levels. During hospitalization, nurses assess patients' needs and provide appropriate care, particularly in areas such as nutrition, elimination, safety, and communication. Nursing interventions aim to ensure patient comfort, safety, dignity, and autonomy.

REFERENCES

1. Roos, R. A. C. (2010). Huntington's disease: a clinical review. Orphanet Journal of Rare Diseases, 5, 40. [Accessed: 07/04/2022]. Available at: https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-5-40#Sec11
2. Mestre, T. A., Shannon, K. (2017). Huntington disease care: From the past to the present, to the future. Parkinsonism & Related Disorders, 44, 114–118. [Accessed: 07/18/2022]. Available at: https://doi.org/10.1016/j.parkreldis.2017.08.009

3. Zielonka, D., Mielcarek, M., & Landwehrmeyer, G. B. (2015). Update on Huntington's disease: advances in care and emerging therapeutic options. Parkinsonism and Related Disorders. [Accessed: 07/18/2022]. Available at: https://www.academia.edu/17269680/Update_on_Huntingtons_disease_Advances_in_care_and_emerging_therapeutic_options?from=cover_page
4. Perkins, A. (2017). Highlighting Huntington disease. Nursing Made Incredibly Easy, 15(4), 28-36. [Accessed: 07/02/2022]. Available at: https://journals.lww.com/nursingmadeincrediblyeasy/Fulltext/2017/07000/Highlighting_Huntington_disease.6.aspx
5. Urrutia, N. L. (2019). Adult-onset Huntington disease. Nursing, 49(7), 36-43. [Accessed: 07/23/2022]. Available at: https://doi.org/10.1097/01.NURSE.0000559914.46449.29
6. Samperi, S., Kwong, P., McGill, T., & Tsui, D. (2021). Huntington's Disease: A Nursing Perspective. Australasian Journal of Neuroscience, 31(2), 18-26. [Accessed: 07/12/2022]. Available at: https://doi.org/10.21307/ajon-2021-007
7. Huntington Disease Society of America. (2014). Caregiver Guide for Mid to Late Stage Huntington's Disease: For Long-Term Care Facilities and In-Home Care Agencies. [Accessed: 07/18/2022]. Available at: http://hdsa.org/wp-content/uploads/2015/04/CaregiverGuide_Mid_Late_StageHD.pdf
8. Huntington's Disease Association. Care in Advanced Huntington's Disease. [Accessed: 07/23/2022]. Available at: https://www.hda.org.uk/media/1171/care-in-advanced-hd-booklet.pdf
9. Greenberg, M., Paddu, N., Bhivandkar, S., & Ahmed, S. (2020). Treating Depression and Suicidality in Huntington's Disease. Psychiatric Annals, 50(2), 85-88. [Accessed: 07/02/2022]. Available at: https://www.academia.edu/download/65109166/treating_depression_and_suicidality_in_huntingtons_disease.pdf
10. Aubeeluck, A., & Wilson, E. (2008). Huntington's disease. Part 1: essential background and management. British Journal of Nursing, 17(3), 146–151. [Accessed: 07/03/2022]. Available at: https://doi.org/10.12968/bjon.2008.17.3.28402
11. Ferraz, C. C. B., Ortega, F. B., Reis, M. G., & Meinberg Cheade, M. (2013). Systematization Of Nursing Care To Patients With Huntington Disease: Case Study. Journal of Nursing UFPE Online, 7(7), 4796-800. [Accessed: 07/23/2022]. Available at: https://periodicos.ufpe.br/revistas/revistaenfermagem/article/viewFile/11734/13972
12. Baker, M., McLaren, S., & Crichton, N. (2016). A review of the literature on nursing in Huntington's disease: A need for specialist knowledge. British Journal of Neuroscience Nursing, 5(8), 360-366. [Accessed: 07/01/2022]. Available at: https://doi.org/10.12968/bjnn.2009.5.8.43595
13. Bates, G. P., Dorsey, R., Gusella, J. F., Hayden, M. R., Kay, C., Leavitt, B. R., et al. (2015). Huntington disease. Nature Reviews Disease Primers. [Accessed: 07/02/2022]. Available at: https://doi.org/10.1038/nrdp.2015.5

ZDRAVSTVENA NJEGA BOLESNIKA OBOLJELOG OD HUNTINGTONOVE BOLESTI

Leonarda-Ana Poljak[1] & Sabina Bis[2]
[1] Karlovačka ljekarna, 47 000 Karlovac-Dubovac, Hrvatska
[2] Veleučilište u Bjelovaru, 43 500 Daruvar, Hrvatska

SAŽETAK

Uvod: Huntingtonova bolest kao progresivno autosomno dominantno neurodegenerativno stanje, okarakterizirano trijadom kliničkih značajki koje utječu na kognitivno, fizičko i psihičko funkcioniranje pojedinca. Obično se simptomi javljaju tokom srednje životne dobi, između 30 i 40 godina, te dovode do smrti jedno do dva desetljeća nakon početka neurološkog pogoršanja. Ova neizlječiva bolest stvara potpunu ovisnost oboljelog o skrbi te značajno utječe na obitelj, prijatelje i bližnje bolesnika. Skrb bolesnika usmjerena je na ublažavanje simptoma, održavanje funkcionalne sposobnosti kao i kvalitete života. Pruža ju multidisciplinarni tim zdravstvenih djelatnika koji osim farmakoloških, uključuju nefarmakološke intervencije.

Cilj rada je istražiti literaturu i cjelovito prikazati proces zdravstvene njege u sestrinskoj skrbi za bolesnika oboljelog od Huntingtonove bolesti.

Zaključak: smjernice za skrb bolesnika oboljelog od Huntingtonove bolesti pružaju temelj za multidisciplinarni pristup i poboljšanje kvalitete života bolesnika, uz naglasak na udobnost, sigurnost i dostojanstvo. Uključivanje specijaliziranih medicinskih sestara, kao ključnih članova tima, doprinosi smanjenju trajanja hospitalizacije, optimizaciji razine skrbi te poboljšanju kvalitete života bolesnika s Huntingtonovom bolesti.

Ključne riječi: bolesnik, Huntingtonova bolest, zdravstvena njega.

Osoba za kontakt: Sabina Bis, sbis@vub.hr