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Royce Copeland

Baylor College of Medicine, royce.copeland30@gmail.com

Shefali Amin

Reading Hospital, shefali.amin@towerhealth.org

Anthony Donato

Reading Hospital - Tower Health, anthony.donato@towerhealth.org

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The Management of Newly Diagnosed Probable Creutzfeldt-Jakob Disease In Acute Rehabilitation Setting: A Case Report

Author ORCID ID:

Royce Copeland <https://orcid.org/0000-0001-9709-5285>

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Creutzfeldt-Jakob disease is a rare and fatal neurodegenerative disease. The purpose of this report is to demonstrate the role of inpatient rehabilitation for a patient recently diagnosed with probable Creutzfeldt-Jakob disease. A 62-year-old male was readmitted to an acute care hospital after a recent mechanical fall. For several weeks, he had a constellation of symptoms, including cognitive impairment and progressive memory loss, gait and postural instability, 20 pounds of unintentional weight loss, visual hallucinations, myoclonus, and hypophonia. After an extensive workup, a diagnosis of probable Creutzfeldt-Jakob disease was made, and he was transferred to an acute inpatient rehabilitation hospital. The patient underwent therapeutic exercise, gait training, neuromuscular reeducation, cognitive behavioral therapy, and voice therapy during his 14-day rehabilitation stay. The patient progressed from moderate assistance to minimal assistance with most activities of daily living, bed mobility, and transfers at the final assessment. He was provided with a rolling walker, home health services, and coordinated care with physicians at discharge.

This case report highlights the unique challenges of individuals and their families who suffer from prion disease and reviews ways to manage these barriers through non-pharmacological and pharmacological treatment options at an inpatient rehabilitation facility. When a diagnosis of probable CJD is made, an inpatient rehabilitation facility can assist in decreasing the caregiver burden and support the psychosocial needs of CJD patients while improving physical functioning.

Keywords

CJD, Creutzfeldt-Jakob disease, prion disease management, rehabilitation, neurodegenerative, dementia

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Conflict of Interest Statement

Conflicts of Interest The authors of this report have no financial interest or funding provided for this project from any source.

CASE REPORT

The Management of Newly Diagnosed Probable Creutzfeldt-Jakob Disease in Acute Rehabilitation Setting: A Case Report

Royce Copeland ^{a,*}, Shefali Amin ^b, Anthony Donato ^b

^a Baylor College of Medicine, Houston TX, USA

^b Reading Hospital/Tower Health, Reading PA USA

Abstract

Creutzfeldt-Jakob disease is a rare and fatal neurodegenerative disease. The purpose of this report is to demonstrate the role of inpatient rehabilitation for a patient recently diagnosed with probable Creutzfeldt-Jakob disease. A 62-year-old male was readmitted to an acute care hospital after a recent mechanical fall. For several weeks, he had a constellation of symptoms, including cognitive impairment and progressive memory loss, gait and postural instability, 20 pounds of unintentional weight loss, visual hallucinations, myoclonus, and hypophonia. After an extensive workup, a diagnosis of probable Creutzfeldt-Jakob disease was made, and he was transferred to an acute inpatient rehabilitation hospital. The patient underwent therapeutic exercise, gait training, neuromuscular reeducation, cognitive behavioral therapy, and voice therapy during his 14-day rehabilitation stay. The patient progressed from moderate assistance to minimal assistance with most activities of daily living, bed mobility, and transfers at the final assessment. He was provided with a rolling walker, home health services, and coordinated care with physicians at discharge.

This case report highlights the unique challenges of individuals and their families who suffer from prion disease and reviews ways to manage these barriers through non-pharmacological and pharmacological treatment options at an inpatient rehabilitation facility. When a diagnosis of probable CJD is made, an inpatient rehabilitation facility can assist in decreasing the caregiver burden and support the psychosocial needs of CJD patients while improving physical functioning.

Keywords: CJD, Creutzfeldt-Jakob disease, Prion disease management, Rehabilitation, Neurodegenerative, Dementia

1. Introduction

Patients with dementia can cause a significant burden on caregivers, resulting in financial stress, depression, physical illness, and decreased quality of life.¹ Creutzfeldt-Jakob disease (CJD), a human prion disease, is clinically characterized by rapidly progressive dementia.² It is believed to be caused by spongiform changes due to the accumulation of the scrapie isoform of the prion protein (PrP^{Sc}) in the brain.² This rare neurodegenerative disorder occurs in just 1 in 1 million people worldwide, or about 350 cases annually in the US.³ A

definitive diagnosis requires a brain biopsy or autopsy.³ With the updated diagnostic criteria, clinicians can make a presumptive premortem diagnosis to allow for planning and goals of care discussions and focus on optimizing function.

The Center for Disease Control has updated diagnostic criteria that divide the diagnosis of CJD into possible, probable, or definite by using a multimodal approach. For a probable diagnosis, a rapidly progressive cognitive impairment must be present with at least two of the following: myoclonus, visual or cerebellar problems, pyramidal or extrapyramidal features, or akinetic mutism. If these symptoms are present along with at least one of the

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* Corresponding author.
E-mail address: royce.copeland@bcm.edu (R. Copeland).

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following: periodic sharp wave complexes on electroencephalogram, hyperintensity signals on diffusion-weighted imaging in the basal ganglia or at least two regions in the cerebral cortex, positive CSF 14-3-3, or positive CSF for real-time quaking-induced conversion (RT-QuIC), then the patient has a probable diagnosis.⁴ A definitive diagnosis can only be made with immunohistochemical or neuropathological brain findings via biopsy at autopsy.⁴ (see [Tables 1 and 2](#))

Although considerable diagnostic advances have been made throughout the years, CJD remains a fatal and non-curable neurodegenerative disease with few therapeutic options. An early presumptive diagnosis is helpful to guide clinicians in supporting families in their decision-making process and transition them into a safe and appropriate discharge.

CJD management is complex and often extends outside the physical domain, and many unique features of prion diseases complicate clinical care. Given the poor prognosis, rapid deterioration, and difficulty in diagnosing this rare condition with limited life expectancy, clinicians may be hesitant to transition CJD patients to inpatient rehabilitation. When time is limited, they must weigh the pros and cons of continuing hospitalization. Therefore, the goal of this report is to help highlight the role inpatient rehabilitation can have on CJD patients and their families.

2. Case presentation

2.1. Hospital summary

A 62-year-old male with no significant past medical history and a recent admission for a mechanical fall resulting in an L1 compression fracture was readmitted to the acute care hospital for rapidly debilitating tremors, visual hallucinations, and a recurrent fall. On more detailed history, the patient was noted to have progressive memory loss with cognitive deficits, 20 pounds of unintentional weight loss, myoclonus, high-frequency low amplitude bilateral hand tremor, hypophonia, hypnagogic visual hallucinations, and postural instability with frequent falls over the past 3–4 months.

Initial admission labs were unremarkable, and a non-contrast CT head did not show acute abnormalities. Given suspicion for CJD, an EEG was performed that showed no epileptiform discharges and specifically no periodic sharp wave complexes that are classic for CJD. Brain MRI found areas of hyperintensity signals within the bilateral occipital and posterior temporal lobes on diffusion-weighted imaging. A lumbar puncture was performed to obtain studies for a paraneoplastic panel, autoimmune encephalitis, and prion disease, all of which were pending when the patient was ready for discharge.

Table 1. Diagnosis of sporadic Creutzfeldt-Jakob disease.

Possible	Probable	Definitive
1. Progressive dementia AND at least two of the following: <ol style="list-style-type: none"> Akinetic mutism Myoclonus Visual or cerebellar signs Pyramidal or extrapyramidal signs 	1. Presence of a neuropsychiatric disorder PLUS positive CSF real time-quaking-induced conversion (RT-QuIC).	Neuropathological confirmation via brain biopsy of any of the following three techniques: <ol style="list-style-type: none"> Immunocyto-chemical Western blot confirmed protease-resistant PrP Presence of scrapie-associated fibrils
AND <ol style="list-style-type: none"> Absence of positive results on CSF RT-QuIC, CSF 14-3-3, EEG, or MRI findings. Duration less than two years Exclusion of other causes in a complete diagnostic workup 	OR <ol style="list-style-type: none"> Rapidly progressive dementia AND at least two of the following: <ol style="list-style-type: none"> Akinetic mutism Myoclonus Visual or cerebellar signs Pyramidal or extrapyramidal signs 	
	AND at least 1 of the following: <ol style="list-style-type: none"> EEG with periodic sharp wave complexes CSF positive 14-3-3 Hyperintensity signaling in the basal ganglia (caudate/putamen) or at least two cortical regions (temporal, parietal, occipital) on diffusion-weighted imaging (DWI) or fluid-attenuated inversion recovery (FLAIR). 	
	AND exclusion of other causes in a complete diagnostic workup	

The table has been adapted from CDC 2018 CJD diagnostic criteria.⁹

Table 2. Nonpharmacologic and pharmacologic interventions for common prion disease symptoms⁶⁻⁸.

Symptom	Management
Myoclonus	Decrease sensory stimulation, maintaining a quiet environment, antiepileptic medications (levetiracetam and sodium valproate), long-acting benzodiazepines (clonazepam and diazepam) if tolerated.
Visual Hallucinations	Cognitive behavioral therapy, family intervention, coping strategies, exposure therapy, acetylcholinesterase inhibitors (donepezil, rivastigmine, galantamine).
Insomnia	Good sleep hygiene, routine sleep and wake schedule, avoid blue light 60-30 min before bedtime. Melatonin agonists, benzodiazepines, and antidepressants with sedating effects (trazodone and mirtazapine).
Psychosis/Aggression/Agitation	Antipsychotic medications (quetiapine, risperidone, or olanzapine).
Depression/Anxiety	Counseling, pastoral care, art, music, and aroma therapy. Antidepressants (SSRI and SNRIs).
Dementia	Pill dispenser, cognitive aids, structured daily routine, offer simple choices, label needed objects, remove clutter or unnecessary objects, use calm and reassuring tone. Acetylcholinesterase inhibitors (donepezil, rivastigmine, galantamine).
Urinary incontinence	Bladder training, fluid schedule, plan toileting schedule, bladder diary, and condom catheter as appropriate.
Constipation	Increase fluid intake, fiber diet, bowel diary, and scheduled toileting.
Dysphagia	Thickened fluids and pureed food. Appropriate positioning with aspiration protocols.
Visual Impairment/Cortical Blindness	Place objects in easy reach of the unaffected side.
Spasticity	Passive movement exercises, massage, acupuncture, botulinum toxin injections, antispasmodic medications (baclofen, dantrolene, tizanidine, benzodiazepines).
Neuropathic Pain	Desensitization techniques (TENS, hot/cold therapy), gabapentin, pregabalin, amitriptyline.
Orthostatic Hypotension	Increase fluid intake, abdominal binder, gradual positioning of body maneuvers, TED stockings, reduce inactivity, midodrine, fludrocortisone, decrease or avoid medications that lower blood pressure, for example (nitrates, diuretics, narcotics, antihypertensive agents, and antiparkinsonian agents).

A probable diagnosis of Creutzfeldt-Jakob disease was made, and given his fall risk and recent reduction in functional status, the patient was transferred to an acute rehabilitation facility to maximize mobility, cognition and coordinate goals of care. Approximately three weeks after obtaining the CSF samples, the CSF results for prion disease were positive for 14-3-3 protein and RT-QuIC, thus confirming a probable diagnosis of Creutzfeldt-Jakob disease with >98% likelihood.

2.2. Acute inpatient rehabilitation summary

Initial assessment revealed the patient to be at moderate assistance for transfers, ambulation, bed mobility, and most activities of daily living. The patient underwent a neurocognitive impairment assessment based on prior education level using the St. Louis University Mental Score and attained a score of 18 out of 30, suggestive of dementia. Using the Boston 6-clicks score to assess basic mobility, the patient scored 12 out of 24, indicating a lower level of independent function. He underwent a comprehensive, multidisciplinary rehabilitation for ambulatory dysfunction, decreased activities of daily living (ADL), strength, postural instability, endurance, and severe cognitive impairment.

Rehabilitation focused on ADL retraining, cognition, gait training, strength, endurance, balance,

safety, family training, and end-of-life care. The patient underwent therapeutic exercise, gait training, neuromuscular reeducation, cognitive behavioral therapy, and voice therapy throughout his 14-day rehabilitation stay. Midodrine, compression stockings, and an abdominal binder managed his significant orthostatic hypotension, and physical therapy worked on his retropulsion with transfers. His inpatient rehabilitation course was complicated by heightened anxiety above his baseline from his new diagnosis, which was managed by escitalopram and cognitive behavioral therapy with a rehabilitation neuropsychologist. Myoclonus was managed by sodium valproate and clonazepam. Educational resources and healthcare provider communication addressed family and patient concerns about diagnosis, transmissibility, and genetic involvement. Palliative medicine focused on realistic expectations, transitioning to end-of-life care, and establishing a living will and power of attorney. He was provided with a rolling walker, home health services, and coordinated care with physicians at discharge.

3. Outcomes/Results

Overall, significant functional gains were obtained at discharge. The patient transitioned from moderate assistance (50% patient effort and 50% caregiver effort) to minimal assistance (75% patient effort and

25% caregiver effort) with most activities of basic daily living, bed mobility, and transfers. The patient was also contact guard assist (100% patient effort and 0% caregiver effort) with a rolling walker for 100 ft. The patient's functional improvements were sufficient to meet goals to return home for a safe discharge.

4. Discussion

Currently, there are no approved treatments for managing the causes of prion disease, and treatment focuses on minimizing symptoms and maintaining the quality of life. One study suggests that the most problematic symptoms of prion disease include coordination and mobility, personal care and incontinence, communication, eating and swallowing, behavior and mood, and cognitive and memory.⁵ These issues can be addressed in the acute rehabilitation unit by a multidisciplinary team. A physiatrist can help the caregiver learn optimal and new approaches for managing the patient's functional and basic needs. This transition of care to a rehabilitation unit can facilitate coordinated care before discharge, arrange for hospice evaluation, counsel families on the end of life and financial matters, and offer social support.⁶ While at the rehabilitation hospital, physiatrists and their healthcare professionals can utilize specific pharmacologic and nonpharmacologic interventions to minimize common and debilitating symptoms that prion disease patients often experience throughout their disease course. The primary focus is to reduce the patient's symptoms and improve quality of life.

The non-pharmacological and pharmacological approaches to treat the varying symptoms one may exhibit with progressive neurodegenerative disease are complex. Myoclonus, orthostatic hypotension, and anxiety were this case's most significant symptomatic medical obstacles. The patient was started on sodium valproate 250 mg daily with clonazepam 0.5 mg at bedtime to combat his myoclonus disturbance. The added effect of moving his room to the end of the rehabilitation unit to decrease sensory stimulation ultimately abated this symptom. Orthostatic hypotension management was a significant barrier in this clinical scenario because it placed safety concerns on the patient's therapy plans. This was ultimately resolved with midodrine 5 mg three times per day, along with an abdominal binder and compression stockings. Lastly, the patient's heightened anxiety about his future and the future of his family's stability was managed with escitalopram 20 mg daily, pastoral/chaplain discussions, and cognitive behavioral therapy with the

neuropsychologist. The combination of pharmacological and non-pharmacological approaches proved to be the best method in this clinical scenario. However, this paper's authors would argue that the most critical component of this patient's rehabilitation was caregiver support, family training, medical education, initiating palliative care, establishing home health services, providing mobility assistive devices, and modifying the living arrangements to meet his functional needs. The patient made substantial improvements in his ability to perform ADLs requiring minimal assistance for ambulation, transfers, bathing, and toileting on discharge. As a result, he and his family successfully managed his care out of their home. The patient passed away six months after discharge from the rehabilitation hospital in his home under the supervision of a hospice team. A subsequent brain autopsy at the National Prion Surveillance Center (www.cjdsurveillance.com) confirmed the presence of the prion protein, thus confirming definite CJD.

There is no research published on outcomes or management of CJD patients in an acute rehabilitation setting, likely due to the rarity of the disease. Therefore, it is unknown when to admit CJD patients appropriately, how long they should be admitted, and how effective rehabilitation can be for those individuals. Suppose this individual was at the end of his disease course. One may argue this individual has limited and valuable time to spend with loved ones, so 14 days of acute rehabilitation would take away from his home experience, and possibly no functional gains would be obtained. This will have to be a clinical decision made by the health provider based on the patient's clinical presentation, family guidance, and patient preference, as there is minimal to no objective data until this case report.

In conclusion, this case report highlights the unique challenges of individuals and their families who suffer from prion disease and reviews ways to manage these barriers through non-pharmacological and pharmacological treatment options at an inpatient rehabilitation facility. A 14-day inpatient rehabilitation stay improved this patient's functional status, trained his caregiving family members, coordinated home health services, and established a relationship with palliative care in anticipation of eventual hospice needs. When a diagnosis of probable CJD is made, an inpatient rehabilitation facility can assist in decreasing the caregiver burden and support the psychosocial needs of CJD patients while improving physical function. Ultimately, the transition of care to an acute rehabilitation hospital can be important to families and individuals affected by prion disease.

Conflict of interest

The authors of this report have no financial interest or funding provided for this project from any source.

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