E-Poster

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Clinical Case: Advanced Neuromotor Rehabilitation in Machado-Joseph Disease With Proximal Tetraparesis

Topic: Miscellaneous

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BACKGROUND

Spinocerebellar ataxia type 3 (SCA3), or Machado-Joseph Disease (MJD), is a hereditary neurodegenerative disorder characterized by progressive ataxia, brainstem dysfunction, and neuromotor impairment. Severe cases can lead to proximal tetraparesis, significantly affecting motor function and quality of life. Machado-Joseph Disease manifests as cerebellar ataxia, loss of coordination in the upper and lower limbs, ophthalmoplegia, and difficulties in speech and swallowing, impacting patient independence. This case presents a rehabilitation approach for a patient with MJD and proximal tetraparesis, integrating neuroplasticity-based strategies, robotic assistance, and intensive multidisciplinary intervention.

CASE DESCRIPTION

Patient: A 62-year-old female, genetically diagnosed with MJD (ATXN3 gene expansion). Symptoms began at age 42, with progressive gait instability. Over the past five years, she developed significant neuromuscular deterioration, mild dysphagia, and dependency in daily activities. Family history includes affected mother and uncle.

Examinations: Brain magnetic resonance imaging (MRI) revealed cerebellar and brainstem atrophy (pons/midbrain). Elektromiyografi (EMG) showed chronic neurogenic patterns with reduced motor potential. Functional assessment: SARA: 29/40 (severe ataxia); FIM: 54/126 (moderate dependency); MRC: 2/5 (proximal tetraparesis); Six-Minute Walk Test (6MWT): 180 m (reduced endurance); Grip strength: 9 kgf (dominant hand).

Rehabilitation Plan: A multidisciplinary rehabilitation program targeted neuroplasticity and mobility. Robotic-assisted gait training improved locomotion, and functional electrical stimulation (FES) enhanced neuromuscular activation. Strengthening exercises focused on proximal muscles to maintain mobility. Occupational therapy addressed fine motor skills and adapted activities for self-care. Balance and coordination training included proprioceptive and weight-shifting exercises. Speech therapy targeted dysphagia management. Fatigue management strategies optimized therapy participation and prevented exhaustion.

Clinical Evolution (2 Months Post-Rehabilitation):

- SARA: 27/40 (improved stability)
- FIM: 60/126 (increased independence)
- MRC: 2+/5 (upper limbs), 3/5 (lower limbs)
- 6MWT: 190 m (enhanced endurance)
- Grip Strength: 10 kgf
- Brain MRI: Mild functional connectivity improvements

DISCUSSION

Although major functional recovery remains limited, intensive multimodal rehabilitation demonstrated improvements in motor control and independence. The integration of assistive technology and targeted interventions proved beneficial in managing the progressive nature of MJD.

CONCLUSION

Despite the progressive nature of MJD, targeted rehabilitation interventions can slow functional decline and help maintain a degree of independence in ADLs. This case reinforces the necessity of realistic rehabilitation goals and multidisciplinary approaches tailored to individual disease progression.

Statement of Disclosures

None

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