PHYSICAL THERAPY AND REHABILITATION OF A PATIENT WITH HEREDITARY SENSORIMOTOR POLYNEUROPATHY CHARCOT-MARIE-TOOTH TYPE 1A - CASE REPORT

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Background

Charcot-Marie-Tooth disease type 1A (CMT1A) is the most common inherited sensorimotor polyneuropathy, caused by a duplication of the PMP22 gene. It is characterized by slowly progressive distal muscle weakness, sensory loss, and foot deformities. Although there is no cure, physical rehabilitation plays a key role in preserving function and alleviating secondary musculoskeletal symptoms. (1,4,6)

Case report

We present a female patient with a genetically confirmed diagnosis of hereditary sensorimotor polyneuropathy CMT1A, initially diagnosed at the National Academy of Sciences of Macedonia six years ago. She reported chronic pain in the neck and right shoulder radiating to the arm with tingling, as well as right hip and buttock pain radiating to the right leg. The patient also experienced longstanding upper limb weakness. Clinical examination revealed preserved cervical lordosis, reduced lumbar lordosis, and increased thoracic kyphosis. Cervical and lumbar spine movements were globally reduced. Both upper extremities showed generalized muscle hypotrophy, particularly in the interosseous muscles of the hands, along with tremor. The range of motion in the shoulders was preserved but painful on the right. Reflexes were symmetrical, and sensory function remained intact. A tailored rehabilitation program was initiated, including physical therapy (pain management and spinal mobility) and kinesitherapy (postural correction, strengthening, and coordination). Progress was assessed using Manual Muscle Testing (MMT), the Timed Up and Go (TUG) test, the 6-Minute Walk Test (6MWT), and the Visual Analog Scale (VAS) for pain. These showed gradual improvement in strength, mobility, and pain

Conclusion

This case illustrates how individualized rehabilitation can improve function and reduce secondary symptoms in CMT1A. Continued therapy is essential to maintaining independence and quality of life in hereditary neuropathies.(2,3)

Keywords: Charcot-Marie-Tooth Disease Rehabilitation Neuropathy Management