

Effectiveness of a multimodal treatment approach for a woman in her 60s with Charcot-Marie-Tooth disease and scoliosis

Abstract

Charcot-Marie-Tooth disease, also known as hereditary peripheral neuropathy, is a rare disorder that causes muscle atrophy and sensory abnormalities. The disease can cause many difficulties in the patient's daily life, especially as muscle atrophy and deformity progresses, making it difficult to walk and maintain balance. This study details the effects of treatment on a woman in her 60s with Charcot-Marie-Tooth disease and severe scoliosis. The treatment approach incorporated a multifaceted approach of increasing skin mobility and range of motion, eliminating catastrophic thoughts, reducing muscle tension, and controlling body weight. During the 3-month treatment period, the patient's primary symptoms, including back pain and joint instability were significantly relieved. In particular, the elimination of catastrophic thoughts and weight control contributed significantly to improving the patient's quality of life. The results of this study suggest the effectiveness of a multimodal treatment approach for the patient with Charcot-Marie-Tooth disease and scoliosis.

Keywords: peripheral neuropathy, muscle atrophy, sensory abnormalities

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Introduction

Charcot-Marie-Tooth disease is a rare disorder known as hereditary peripheral neuropathy that causes muscle atrophy, weakness, and sensory abnormalities, primarily in the lower extremities. The disease affects nerve transmission and the patients may experience many difficulties in their daily lives. In particular, progressive atrophy and deformity of the leg muscles often result in gait disturbance and balance problems. Below is an overview of Charcot-Marie-Tooth Disease.¹

Charcot-Marie-Tooth disease (CMT)

i. Definition

Charcot-Marie-Tooth disease (CMT) is an inherited peripheral neuropathy that causes abnormal peripheral nerve structure and function, resulting in muscle atrophy and loss of sensation.

ii. Disease classification

CMT is classified into several subtypes based on the location and type of gene mutation.

CMT1: Primarily due to abnormalities in the myelin sheath, it causes a slowing of nerve conduction velocity.

CMT2: Mainly due to abnormalities in the nerve fibers themselves, it causes nerve conduction velocity is within normal limits.

CMT3: Severe form that develops in infancy. It is characterized by demyelination.

CMT4: Autoimmune form with inherited genetic mutations from both parents.

iii. Symptoms

Motor Symptoms: Atrophy and weakness of the leg muscles are often the initial signs. As the disease progresses, hand muscles may also be affected.

Sensory symptoms: loss of sensation or abnormal sensations beginning at the tips of the fingers of the feet and hands.

Foot deformities: foot deformities such as high plantar feet and hammertoes may be present.

Diagnosis: CMT can be diagnosed by the following criteria

iv. Family history

Since CMT is a genetic disorder, check for the presence of symptoms in family members.

Nerve conduction velocity studies: measure the speed of nerve transmission.

Electromyography: Evaluates the electrical activity of muscles.

Genetic testing: to identify specific genetic mutations.²

v. Treatment

Rehabilitation: Physical therapy to maintain or increase muscle strength and range of motion of joints.

Orthotics: Orthotics to correct foot deformities and difficulty walking.

Medications: Medications to relieve pain and muscle spasms.

Prognosis: CMT is a progressive disease.

Although CMT is a progressive disease, in most cases it does not affect life expectancy. However, progression of symptoms may cause difficulty with activities of daily living and walking.

Due to its genetic diversity and range of symptoms, Charcot-Marie-Tooth disease often requires different characteristics and treatment approaches for each patient. In this case report, we describe the clinical course and treatment details of a woman in her 60s who underwent five times spinal fusion surgeries against a background of Charcot-Marie-Tooth disease.

Summarize the rehabilitation of a typical CMT

i. Physical therapy

Strength training: Strength training with light resistance is used to strengthen atrophied muscles.

Stretching: Regular stretching is recommended to maintain flexibility of muscles and tendons.

Balance training: Exercises to improve balance and coordination are performed to decrease the risk of falling.

Gait training: Patients are trained to learn more efficient and safer gait patterns while using orthotics.

ii. Occupational therapy

Activities of Daily Living (ADL) Training: Strategies are taught to improve the efficiency of activities of daily living such as eating, dressing, and writing.

Use of adaptive equipment: Learn to use special tools and devices to facilitate activities of daily living.

iii. Orthotics

Foot Orthotics (AFOs): used to keep the feet properly positioned and to stabilize gait.

Wrist Orthotics: may be used to assist with hand function and facilitate activities of daily living.

iv. Pain management

Heat/cool therapy: used to reduce pain and inflammation.

Electrical stimulation therapy: may be used to manage pain and improve muscle function.

v. Exercise therapy

Hydrotherapy: uses the buoyancy of water to reduce stress on the joints while exercising.

Bicycle exercises: low-impact exercises that help maintain muscle strength.

vi. Lifestyle changes

Weight control: Maintaining an appropriate weight is recommended, as being overweight puts extra stress on the legs and joints.

Nutritional guidance: A healthy diet is important to support overall health and muscle function.

Case

i. Patient information

Age: 60s

Gender: Female

Height and weight: 144cm, 48kg

Occupation: Office Work

ii. Main complaint

Backache, Right hip weakness, discomfort in both ankles, scoliosis.

iii. Previous medical history

Charcot-Marie-Tooth disease, five times' spinal fusion surgeries were performed between February to June 2021 (Figures 1&2).



Figure 1 Pre operation.



Figure 2 Post operation.

iv. Current medical history

The patient has Charcot-Marie-Tooth disease as a congenital condition. She also has scoliosis and underwent spinal fusion surgery in 2021. Her chief complaint is back pain after spinal fusion surgery and radiating pain from Th3 to Th12 (NRS score 10) with instability and occasional motor pain when standing up and going up and down stairs. She has hip joint instability and uses orthopedic shoes for her lower extremity apex foot.³

v. Diagnosis

Charcot-Marie-Tooth disease, scoliosis

vi. Treatment and intervention

1. Skin stretching to produce the back skin glide

Stretching and acupressure are used to soften skin that has become hard due to decreased skin movement, thereby increasing mobility and range of motion, as well as promoting cutaneous sensation and positional perception and activating muscle activity. As a result of promoting muscle activity, pain substances are eliminated and pain is suppressed.

2. Psychotherapy to prevent catastrophic thinking

The patient needed to rewrite his perception of pain as normal due to chronic pain over a period of more than three years. Cognitive-behavioral therapy was used to decrease the amount of time spent feeling pain in daily life by decreasing the number of behaviors that cause strong pain. In addition, to maintain a positive psychological state, he made it a habit to guide himself with affirmations.

3. Promoting muscle activity utilizing neuromuscular stimulation techniques

PNF was performed as exercise rehabilitation of the hip joint. Resistance exercise was performed in the final range of motion, and gradually shifted to assisted exercise for mobilization. Aroma oil was used to stimulate the parasympathetic nervous system to relieve muscle tension and promote the efficiency of hip flexion and extension muscles.⁴

4. Weight loss due to dietary restriction

Charcot-Marie-Tooth disease often causes obesity in adolescence and beyond due to exercise restrictions. The present patient weighed 48 kg at the beginning of treatment and achieved a weight loss of 8 kg in 2 months with dietary guidance.

vii. Progress and results

In the third month of treatment (4 sessions), back pain NRS10 decreased to 3, one-leg standing balance test improved from 1 second to 10 seconds, and hip stability improved. Weight decreased from 48 kg to 40 kg and hip pain was also reduced. In addition, the reduction in pain eliminated catastrophic thinking, which in turn eliminated anxiety about future life and work.

Consideration

Although this patient has severe scoliosis associated with Charcot-Marie-Tooth disease, a multifaceted approach that included skin movement, increased joint range of motion, elimination of catastrophic thinking, relaxation of muscle tone, and weight control may have resulted in symptomatic relief at this early stage. In this case, it is unclear whether weight loss had a direct impact on the improvement in pain. Since Charcot-Marie-Tooth disease causes progressive muscle atrophy, it is unclear whether the weight loss due to dietary restriction was due to a decrease in body fat or a decrease in muscle mass. This is an issue to be addressed in the future.⁵

Conclusion

A multifaceted motor, psychological, and nutritional approach was effective for the patients with Charcot-Marie-Tooth disease and severe scoliosis.

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None.

Conflicts of interest

The author declares that there are no conflicts of interest.

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