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Physical Therapy and Rehabilitation of a Patient with Hereditary Sensorimotor Polyneuropathy Charcot-Marie-Tooth Type 1A - Case Report

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Abstract

Charcot-Marie-Tooth disease type 1A (CMT1A) is the most common hereditary demyelinating peripheral neuropathy, characterized by progressive distal muscle weakness, sensory loss, and functional impairment. Despite its prevalence, evidence-based rehabilitation protocols remain limited, particularly for adults with long-standing disease. This case report presents a 49-year-old female with genetically confirmed CMT1A who completed a 20-day individualized rehabilitation program combining physical therapy and kinesitherapy. Pre- and post-intervention 2 assessments included Manual Muscle Testing (MMT), Timed Up and Go (TUG), 6-Minute Walk Test (6MWT), and Visual Analog Scale (VAS) for pain. Following treatment, the patient demonstrated measurable improvements: upper and lower limb strength increased by 0.5 MMT grades, TUG time decreased by 2.4 seconds, 6MWT distance increased by 40 meters, and pain reduced from 6/10 to 3/10. These results highlight the potential of intensive, short-term, multimodal rehabilitation to improve muscle strength, balance, endurance, and pain in CMT1A patients.

Keywords: Charcot-Marie-Tooth Disease, Cmt1a, Hereditary Sensorimotor Polyneuropathy, Physical Therapy, Kinesitherapy, Rehabilitation, Muscle Strength.

Introduction

Charcot-Marie-Tooth disease (CMT) is a heterogeneous group of inherited peripheral neuropathies characterized by progressive degeneration of the peripheral nerves, affecting both motor and sensory function. Among its subtypes, Charcot-Marie-Tooth type 1A (CMT1A) is the most prevalent, accounting for approximately 60–70% of demyelinating CMT cases worldwide [1, 2]. CMT1A is caused by a duplication of the peripheral myelin protein 22 (PMP22) gene on chromosome 17p11.2, leading to abnormal myelin formation, slowed nerve conduction, and distal axonal degeneration [3, 4]. This genetic abnormality results in progressive distal muscle weakness, sensory deficits, and characteristic deformities, such as pes cavus, claw toes, and hand muscle atrophy [2].

The prevalence of CMT1A has been estimated at 1 in 5,000 individuals globally, with clinical manifestations typically appearing during childhood or adolescence, although late-onset cases have been reported [3, 5]. Disease progression is gradual, and patients often experience functional limitations in daily activities, including walking, stair climbing, and fine motor tasks such as writing or buttoning [4]. Fatigue, frequent tripping, and falls are common, contributing to reduced independence and quality of life [6]. Upper limb involvement often occurs later in the dis-

ease course, further impairing manual dexterity and increasing dependence on others for activities of daily living [2]. Chronic musculoskeletal pain is frequently reported, originating from muscle imbalance, joint deformities, and compensatory postures [7].

Pathophysiology of CMT1A involves the overexpression of PMP22, which disrupts the myelin sheath, leading to demyelination, slowed nerve conduction, and secondary axonal loss [1]. The distal muscles are typically affected first, as longer axons are more susceptible to demyelination-related degeneration. Sensory deficits primarily involve vibration and proprioception, while motor deficits manifest as distal weakness and atrophy. Compensatory strategies, such as high-stepping gait and wrist extensor recruitment, often develop to maintain mobility but may increase energy expenditure and fatigue [4].

Early diagnosis is critical for planning effective management strategies. Genetic testing enables precise confirmation of CMT1A and informs prognostic expectations [3]. While no curative treatment exists, supportive and rehabilitative interventions are the cornerstone of management. These include orthotics, physical therapy, occupational therapy, exercise programs, and patient education to prevent secondary complications and maintain independence [8].

Rehabilitation for CMT1A focuses on improving muscle strength, joint mobility, coordination, balance, and functional endurance. Evidence indicates that structured programs combining strengthening, balance training, functional mobility exercises, and manual therapy can improve both objective functional outcomes and patient-reported quality of life [6, 4]. Resistance training targeting distal and proximal muscles has been associated with improved manual dexterity, walking distance, and reduced fatigue, while task-specific functional exercises enhance independence in daily activities. Pain management through postural correction, stretching, and manual therapy also supports active participation in rehabilitation and reduces musculoskeletal discomfort [7].

Despite growing evidence, there is a lack of high-quality case reports documenting short-term, intensive rehabilitation programs in adults with long-standing CMT1A. This report presents a 49-year-old female patient who underwent a 20-day tailored program integrating physical therapy and kinesitherapy, with pre- and post-intervention assessments using validated functional measures, including Manual Muscle Testing (MMT), Timed Up and Go (TUG), 6-Minute Walk Test (6MWT), and Visual Analog Scale (VAS) for pain. The case aims to illustrate the potential for short-term, individualized interventions to produce meaningful functional improvements and guide clinical practice for hereditary neuropathies.

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Methodology (Case Presentation & Intervention)

Patient Presentation

The patient was a 49-year-old female with genetically confirmed Charcot-Marie-Tooth disease type 1A (CMT1A), diagnosed six years prior through molecular genetic testing identifying a duplication of the PMP22 gene [3]. She presented with progressive distal muscle weakness, upper limb weakness affecting daily activities, chronic pain in the neck, right shoulder, and right hip, and paresthesia radiating along the right upper and lower limbs. She reported frequent tripping, difficulty with stairs, and fatigue with prolonged walking or standing. Family history revealed no known relatives with CMT1A, suggesting a de novo mutation. The patient had no significant comorbidities and no prior orthopedic surgeries.

Clinical Examination

A detailed assessment of posture, musculoskeletal function, and neurological status was performed:

- Posture: Preserved cervical lordosis, reduced lumbar lordosis, increased thoracic kyphosis. Mild anterior pelvic tilt observed.
- Cervical and Lumbar Spine Mobility: Globally reduced; pain present on right lateral flexion and rotation.
- Upper Limb: Generalized muscle hypotrophy, particularly in interosseous hand muscles; mild hand tremor; reduced grip strength (MMT 3/5). Shoulder ROM preserved but painful on right.
- Lower Limb: Distal weakness (MMT 3.5/5), mild pes cavus deformities; preserved patellar and Achilles reflexes; sensation intact.
- Balance and Gait: Mild instability during tandem gait; tendency to overstep during walking; compensated high-stepping gait noted.
- Pain Assessment: Visual Analog Scale (VAS) score: 6/10.

Rehabilitation Program

The patient underwent a 20-day intensive rehabilitation pro-

gram, combining physical therapy and kinesitherapy, with individualized progression based on tolerance and functional goals.

1. **Physical Therapy**

- Manual therapy for cervical, thoracic, and lumbar spine to improve mobility and reduce musculoskeletal strain [4].
- Stretching exercises target hamstrings, calf muscles, and paraspinal musculature to alleviate tightness and postural imbalances.
- Pain management using Transcutaneous Electrical Nerve Stimulation (TENS), thermotherapy, and guided posture correction.
- Education on proper ergonomics and postural alignment during daily activities.

2. **Kinesitherapy**

- **Strengthening Exercises:**
- Upper limb: resistance band exercises for interosseous muscles, wrist flexors/extensors, deltoids (3 sets × 10 reps).
- Lower limb: ankle dorsiflexion, plantar flexion, quadriceps, and gluteal strengthening (3 sets × 12 reps).
- **Coordination and Balance Training:**
- Tandem walking, single-leg stance, obstacle negotiation, and step-ups to enhance proprioception and postural stability.
- Fine motor training: grasp and release tasks, buttoning, writing, and small object manipulation to improve dexterity.
- **Functional Mobility:**
- Sit-to-stand repetitions, stair climbing, and gait training over variable surfaces.
- Treadmill walking with progressive duration and intensity.
- Sessions were two times daily, 60 minutes each, with intensity adjusted according to fatigue, pain, and tolerance.

Table 1: Daily Rehabilitation Schedule during the 20-Day Program

Day	Morning Session (60 min)	Afternoon Session (60 min)	Notes
1–5	Stretching + Manual Therapy + Balance	Strengthening + Gait Training	Pain <5/10
6–10	Stretching + Postural Correction	Strengthening + Functional ADL tasks	Slight fatigue
11–15	Stretching + Manual Therapy	Resistance training + Coordination exercises	Monitor VAS
16–20	Stretching + Postural + Balance	Functional mobility + Endurance training	Progressive load

Outcome Measures and Assessment Tools

Functional improvements were measured before and after the 20-day program using validated clinical tools:

- Manual Muscle Testing (MMT): Assessed proximal and distal muscle groups in both upper and lower limbs to monitor strength changes.
- Timed Up and Go (TUG) Test: Evaluated dynamic balance, mobility, and fall risk (Shy et al., 2018).
- 6-Minute Walk Test (6MWT): Measured functional walking endurance (Ridolfi et al., 2017).
- Visual Analog Scale (VAS): Quantified patient-reported pain intensity, allowing tracking of analgesic effects of rehabilitation interventions.

Progress Monitoring and Program Adaptation

Daily monitoring of pain, fatigue, and performance allowed real-time adjustments in exercise intensity, repetitions, and functional tasks. Exercises were progressed gradually to prevent overuse injuries and optimize neuromuscular adaptation. Compliance was high, and no adverse events occurred. The patient also received educational counseling to encourage home exercises post-discharge, ensuring continuity of care.

Rationale for Intervention

The combination of physical therapy and kinesitherapy was designed to:

- Improve distal and proximal muscle strength, particularly in muscles affected by demyelination and atrophy.
- Enhance balance and coordination, reducing risk of falls and improving gait efficiency.
- Increase functional mobility through repetitive, task-specific practice.
- Alleviate chronic musculoskeletal pain via manual therapy, stretching, and posture correction.
- Promote patient engagement and self-efficacy, essential for adherence and long-term functional maintenance [4, 6].

The integration of objective functional measures allowed precise quantification of outcomes, guiding ongoing therapy and providing evidence of program efficacy.

Results and Discussion

Functional Outcomes

Following the 20-day individualized rehabilitation program, the patient demonstrated measurable improvements across all functional and pain assessment measures. These improvements provide evidence of the effectiveness of a short-term, intensive, tailored rehabilitation intervention in a patient with long-standing CMT1A.

Manual Muscle Testing (MMT): Upper limb strength improved from an average of 3.0/5 to 3.5/5, while lower limb strength increased from 3.5/5 to 4.0/5. These gains reflect enhanced neuromuscular recruitment, distal and proximal muscle performance, and overall upper and lower extremity function. Targeted resistance exercises likely contributed to these improvements, in agreement with previous studies demonstrating that progressive resistance training enhances muscle strength and hand dexterity in CMT patients [7, 4].

Timed Up and Go (TUG) Test: The patient's TUG time decreased from 16.2 seconds to 13.8 seconds, indicating improved dynamic balance, mobility, and fall risk reduction. Balance and coordination exercises, such as tandem walking and single-leg stance, along with functional task training, likely facilitated better postural control and faster transitions from sitting to standing. These findings align with the work of Shy et al. (2018), who reported that task-specific balance training improves mobility and reduces fall risk in CMT populations.

6-Minute Walk Test (6MWT): Walking distance increased from 320 meters to 360 meters, demonstrating enhanced endurance, gait efficiency, and cardiovascular conditioning. The progressive lower limb strengthening, gait training, and treadmill exercises likely contributed to improved energy expenditure and functional walking capacity. This is consistent with Ridolfi et al. (2017), who highlighted that structured exercise programs improve walking distance in hereditary neuropathies.

Visual Analog Scale (VAS) for Pain: Pain intensity decreased from 6/10 to 3/10, reflecting effective pain management through manual therapy, postural correction, and gradual mobilization. Reductions in musculoskeletal pain likely facilitated increased participation in therapy sessions, promoting further functional gains. Similar effects have been observed in prior studies where individualized rehabilitation led to pain reduction and improved patient satisfaction [7].

Table 2: Functional Assessment Before and After Therapy

Test	Baseline	Post-Therapy	Change	Interpretation
MMT (upper limb avg)	3.0	3.5	+0.5	Moderate improvement in upper limb muscle strength.
MMT (lower limb avg)	3.5	4.0	+0.5	Moderate improvement in lower limb muscle strength.
TUG (seconds)	16.2	13.8	−2.4	Faster time indicates better mobility and functional performance.
6MWT (meters)	320	360	+40	Greater distance walked indicates improved endurance.
VAS (pain)	6	3	−3	Pain reduction suggests effective symptom relief.

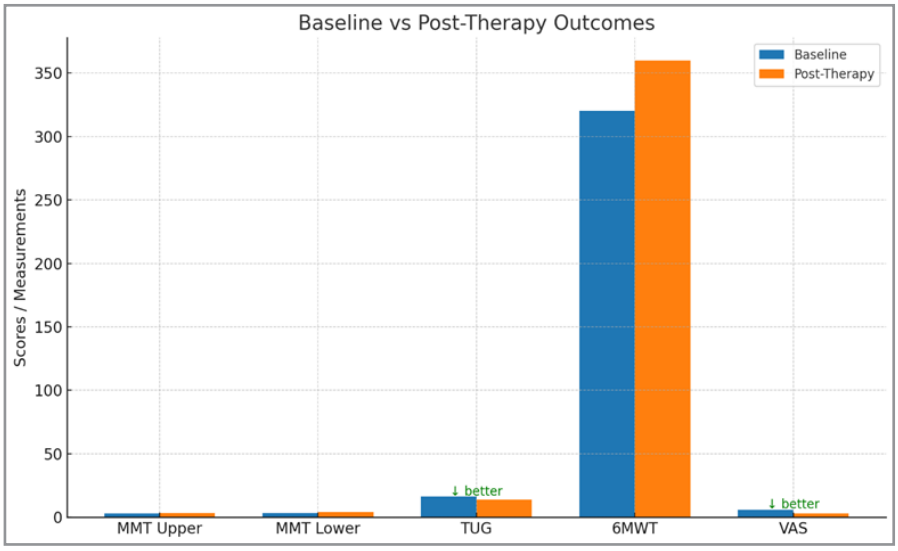

Figure 1: Functional Improvement Graph

Discussion

The observed functional improvements highlight the benefits of individualized rehabilitation in adult patients with long-standing CMT1A. Resistance exercises targeting both distal and proximal muscles enhanced muscle strength, contributing to improved MMT scores. Balance and coordination training promoted faster TUG times and safer gait patterns, while endurance-focused activities such as treadmill walking increased 6MWT distance. Pain reduction through manual therapy and posture correction improved overall participation and adherence, underscoring the interplay between functional and symptomatic outcomes.

These results are consistent with previous literature supporting multimodal rehabilitation approaches for CMT patients. Ridolfi et al. (2017) demonstrated that structured physical therapy improves muscle strength, walking endurance, and functional independence. Shy et al. (2018) emphasized that task-specific training and balance exercises enhance mobility and reduce fall risk. Furthermore, individualized programs addressing specific deficits, patient tolerance, and progression are more effective than generic exercise protocols [7].

Clinical Implications

The case illustrates that even short-term, intensive interventions can produce meaningful functional gains in adult CMT1A patients. Clinicians should consider:

- Tailoring exercise programs based on the patient's muscle strength, balance, and functional deficits.
- Incorporating multimodal therapy: strengthening, balance, coordination, functional mobility, and pain management.
- Monitoring patient tolerance, fatigue, and pain to adjust intensity safely.
- Using objective outcome measures (MMT, TUG, 6MWT, VAS) for tracking progress and guiding therapy modifications.

Limitations

This case report is limited by its single-patient design and short

intervention period. CMT1A is a progressive condition, so long-term outcomes and sustainability of improvements are uncertain. Larger studies and randomized controlled trials are needed to establish evidence-based protocols for optimal rehabilitation duration, intensity, and combination of modalities.

Future Directions

Future research should be investigated:

- Long-term effects of individualized rehabilitation in CMT1A.
- Comparative efficacy of different exercise modalities (resistance vs. aerobic vs. functional training).
- Integration of technology-assisted therapy, such as robotics or virtual reality, to enhance adherence and engagement.
- Patient-reported outcomes on quality of life, fatigue, and participation in daily activities.

In conclusion, this case reinforces that tailored, evidence-based rehabilitation programs can significantly improve functional capacity, reduce pain, and enhance quality of life in patients with CMT1A, even over a short intervention period.

Conclusion

This case report demonstrates that a short-term, intensive, individualized rehabilitation program can yield significant functional improvements in a patient with genetically confirmed Charcot-Marie-Tooth type 1A (CMT1A). Over a 20-day intervention, the patient achieved measurable gains in muscle strength, balance, functional mobility, endurance, and pain reduction, as evidenced by improvements in MMT, TUG, 6MWT, and VAS scores.

The observed improvements highlight the importance of personalized rehabilitation protocols tailored to the patient's specific deficits and tolerance. Strengthening exercises effectively enhanced distal and proximal muscle function, coordination and balance training facilitated safer and more efficient mobility, and functional task practice contributed to greater independence in daily activities. Pain management strategies, including manual therapy and postural correction, played a key role in improving participation and adherence, enabling maximal benefit from therapy [7, 4].

This report underscores that even short-term interventions can be clinically meaningful, suggesting that structured rehabilitation should be considered an integral component of CMT1A management. Continued therapy is essential to maintain these gains over time, given the progressive nature of hereditary neuropathies. The use of objective, validated outcome measures allow clinicians to monitor progress, guide therapy adjustments, and provide tangible evidence of functional improvement, thereby enhancing patient motivation and engagement.

Moreover, this case highlights the potential for rehabilitation to improve quality of life, not only through physical improvements but also by reducing pain and promoting self-efficacy in daily activities. While the findings are limited by single-patient design and short intervention period, they provide valuable insights into practical, evidence-based rehabilitation strategies for adults with long-standing CMT1A.

In conclusion, the case contributes to the growing body of evidence supporting tailored, multimodal rehabilitation programs as a cornerstone in the management of CMT1A. Future studies should explore the long-term sustainability of functional gains, optimal program duration and intensity, and integration of innovative therapeutic modalities to further enhance outcomes in this population [6, 4].

Ethical Considerations

This case report was conducted in accordance with the Declaration of Helsinki and national ethical guidelines, ensuring the protection of patient rights, safety, and confidentiality throughout the study.

Informed Consent

Written informed consent was obtained from the patient prior to the initiation of the rehabilitation program and for the publication of this case report. The patient received detailed information about the purpose, procedures, expected benefits, potential risks, and voluntary nature of participation.

Confidentiality

All patient-identifying information was anonymized. Tables, figures, and textual descriptions were carefully de-identified to maintain the patient's privacy.

Ethical Approval

The study protocol was reviewed and approved by the Institutional Ethics Committee of the University Clinic for Physical Medicine and Rehabilitation, Faculty of Medicine, "St. Cyril and Methodius," Republic of North Macedonia.

Conflict of Interest

The authors declare no potential conflicts of interest regarding the research, authorship, or publication of this article.

Funding

No external funding or financial support was received for this study. The rehabilitation program was conducted as part of routine clinical care.

Safety and Compliance

Patient safety was continuously monitored throughout the 20-day rehabilitation program. Adjustments to exercise intensity, repetitions, and functional tasks were made daily based on fatigue, pain, and tolerance. No adverse events, injuries, or complications were reported. Compliance with the therapy program was high, and patient engagement was supported through education on home exercises and functional mobility strategies.

Ethical Justification for Publication

The publication of this case provides valuable insight into the effectiveness of individualized rehabilitation in adults with long-standing CMT1A, contributing to the limited literature in this area while ensuring the patient's anonymity and rights were fully protected.

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