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PROPOSAL OF PHYSICAL EXERCISES FOR CHARCOT MARIE TOOTH DISEASE PATIENTS: A TAILORED APPROACH

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A. Study design/planning
B. Data collection/entry
C. Data analysis/statistics
D. Data interpretation
E. Preparation of manuscript
F. Literature analysis/search
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Abstract:

Background: Charcot-Marie-Tooth disease (CMT) is a neurological condition of genetic etiology classified among rare diseases. The aim of the study was to subject two CMT-1A patient sisters to an ad-hoc training program of 6 weeks (18 sessions) and to record and obtain improvements in proprioception, stability, balance, mobility, and strength.

Methods: Two sisters aged 32 and 30 respectively, with moderate disability (CMT-Neuropathy Score (CMTNS) of 11/36), with pes cavus, hindfoot varus, and a limited ankle range of motion in foot dorsiflexion. General weakness, early fatigue, and impaired gait were also present in the two patients. Both patients, with the same initial deficits, underwent this new intense training program consisting of 60-minute workouts 3 times a week any other day for 6 weeks (a total of 18 sessions).

Results: The study showed positive results on the following scales: Romberg test; 6 MWT; 30-ACT; 30 CST; TUG; Q-Walk-12, and Q-SF-36.

Conclusion: The study aimed to underline the importance of an early disease training approach to limit its progression. The study demonstrated a positive impact of a 6-week gym training program based on monopodalic standing and quadrupedal exercises on proprioceptive abilities, postural stability, and improved balance.

Introduction

Charcot-Marie-Tooth disease (CMT) is a neurological disease of the peripheral nervous system (PNS), with a genetic etiology that affects 1 in every 2,500 people [1]. From a clinical point of view, it is a sensory-motor, length-dependent, slowly progressive polyneuropathy characterized by symmetrical distal weakness of the lower limbs, as well as tactile hypoesthesia, gradually worsening proximally and distally, and causing an alteration of the feet, which become cavus-shaped and floppy [2, 3]. Over time, patients are forced to raise their knees more than normal to avoid tripping with their toes: this gait, which resembles that of the horse, is called "stepping" or equine. Weakness in the upper limbs mainly involves the hands, with particular difficulty in buttoning up and unbuttoning shirts, opening a bottle, and turning the keys in the lock. There is typically a reduction or absence of deep tendon reflexes [4] and muscle atrophy. Distal upper limb tremor, muscle cramps (especially in the feet and legs), acrocyanosis, and cold feet may also occur, and rarely, there may also be central nervous system impairment [5]. In most patients, Charcot-Marie-Tooth disease does not affect life expectancy, but significantly worsens the quality of life and reduces motor autonomy, especially in older adulthood [6].

The age of onset of the disease varies but in most cases, the first signs appear at the end of the first decade or during the second decade of life, with a chronic and slowly progressive evolution. It is well known that in healthy individuals, exercise has countless health benefits, including a reduced risk of obesity, osteoporosis, heart disease, and diabetes [7]. Exercise for people with Charcot-Marie-Tooth (CMT) disease can help maintain strength and function, especially using muscle-strengthening exercises, stretching, and functional exercises. Wallace et al. found that for people with CMT, aerobic training in community gyms was safe and improved aerobic capacity [8]. The purpose of this research is to propose a new training program with the goal of achieving motor, functional, and balance improvement in CMT patients. For the first time, this new training was applied to two sisters affected by CMT-1A.

Methods

E. and J. are two sisters aged 32 and 30 respectively, with moderate disability (CMT-Neuropathy Score (CMTNS) of 11/36), with pes cavus, hindfoot varus, and a limited ankle range of motion in foot dorsiflexion. General weakness, early fatigue, and impaired gait were also present in the two patients. Both patients, with the same initial deficits, underwent this new intense training program consisting of 60-minute workouts 3 times a week any other day for 6 weeks (a total of 18 sessions). The training protocol focused on improving muscle strength of the lower and upper limbs, mobility, and stretching. The participants were subjected to 7 tests: Romberg test; Six Minute Walk Test (6 MWT); 30-Arm Curl Test (ACT); 30-Chair Standing Test (CST); Time Up and Go (TUG); Twelve Item Walking Scale Questionnaire (Walk-12) and Quality of Life Assessment Questionnaire (SF-36). The improvement was defined as the difference between T1 (after 6 intensive weeks of training) and T0 (before training) split into 4 degrees of improvement: 0 (T1-T0 <5%); 1 (T1-T0 5-10%); 2 (T1-T0 10-15%); 3 (T1-T0 15-20%) and 4 (T1-T0 >20%).

Results

The proposed training exercises and the results obtained from the tests before and after the training are presented in Table 1.

Table 1. Training program followed by the participants in the gym under the supervision of a neurologist and a clinical kinesiologist and the results obtained in the various tests at baseline (T0) and after 6 weeks of training (T1)

TRAINING PHASES	3 SESSIONS A WEEK ANY OTHER DAY	EXERCISE	REPETITION / SET
INITIAL Session duration 15′	Lower/upper limb mobility Neck, shoulders, pelvis and ankles	– Circumduction – Passive tibio-tarsal dorsiflexion with elastic band – Achilles tendon stretching	30″ for 3 sets each exercise
CENTRAL Lower limbs Session duration 20′	Strengthening of the tibial and peroneal muscles	In active isometrics, maintaining dorsiflexion in supine decubitus position and while seated	10″ each side x 10 times x 3 series
	Glutes, quadriceps and hamstrings	Squat-free and with fitball Hip thrusts Leg extensions Leg curls	3x12 3x10 3x12 3x12
Upper limbs Session duration 15′			
	Strengthening the flexors and extensors of the elbow and shoulders	Dumbbell curls Pushdowns/double kickbacks Lateral raises	3x16 3x12 3x12
FINAL Session duration 10′	Abdomen ed equilibrium	Plank Alternate lunges Contralateral limb extension in quadrupedal position	3x 30″ 3x10 3x10

TEST	PATIENT, T_0 [1]	PATIENT, T_1 [2]	PT. IMPROVEMENT (%) ; (0-4)[3]
Romberg Test	E. Positive	E. Positive	0
	J. Negative	J. Negative	0
Six-Minute Walking Test (6 MWT)	E. 538 m	E. 546	E. 1,5% ; 0
	J. 517 m	J. 562	J. 8,7% ; 1
30-Arm Curl Test (ACT)	E. 21[4]	E. 27	E. 28,6% ; 4
	J. 21[4]	J. 29	J. 38,1% ; 4
30-Chair Stand Test (CST)	E. 21[4]	E. 25	E. 19,0% ; 3
	J. 20[4]	J. 30	J. 50,0% ; 4
Time Up and Go (TUG)	E. 5.36″	E. 5.38″	E. - 0,4% ; 0
	J. 5.85″	J.5.40″	J. 7,7% ; 1
Twelve Item Walking Scale Questionnaire (Walk-12)	E. 22	E. 22	E. 0% ; 0
	J. 25	J. 23	J. 8,0% ; 1
Assessment of Quality of Life Questionnaire (SF-36)	E. 54,86	E. 55,70	E. 1,5 % ; 0
	J. 71.52	J. 75	J. 4.9 % ; 0

[1] T0 represents the patient's capabilities before starting training

[2] T1 represents the patient's capabilities after successfully completing the 6-week intensive training program.

[3] The improvement was defined as the difference between T1 and T0 split in 4 categories: 0 (T1-T0 <5%); 1 (T1-T0 5-10%); 2 (T1-T0 10-15%); 3 (T1-T0 15-20%) and 4 (T1-T0 >20%).

[4] Repetitions

Figure 1 illustrates the improved ability to perform a deeper squat without heel-lifting compensation obtained following the exercises designed to reduce the level of muscle recruitment between the anterior and posterior muscle fibers of the distal part of the lower limb. The training program applied in this study allowed patients to achieve improvements in both mobility and strength.

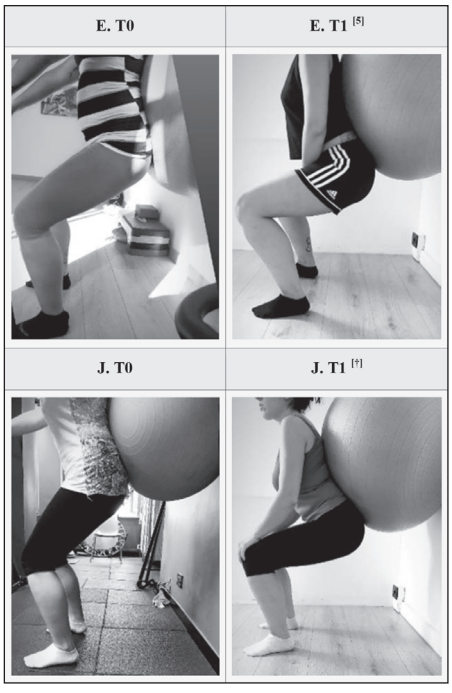

Figure 1. Illustration of mobility of the ankle joints before and after the 6-week intensive training program

[5] Improved ability to perform a deeper squat without heel-lifting compensation after successful completion of the training program

Discussion

In the scientific community, exercise has been commonly discouraged in neuromuscular diseases, primarily due to theoretical considerations rather than adverse outcomes [9]. Without supporting data, it has been postulated that a weak muscle is more susceptible to damage due to overwork because it is already functioning close to its maximal capabilities [10]. This study aimed to demonstrate that a 6-week gym training program using controlled exercises has a positive impact on proprioceptive abilities, in particular the perception of the body position in space and the state of contraction of muscles. It also improved postural stability, by restoring the balance between the accentuated retraction of the Achilles tendon and the capacity of the ankle dorsiflexor and pronator muscles, by focusing on stretching these structures. Moreover, this study showed balance improvements with monopodalic standing and quadrupedal exercises.

This study is not without limitations. Both patients had various osteoskeletal abnormalities, such as pes cavus, hindfoot varus, tibiotarsal reduced range of motion, and callosities at the head of the 1st and 5th metatarsals. Furthermore, patient E. complained of generalized quadriceps cramps during the first training sessions, while J. developed some lower extremity edemas which subsequently resolved. These may have been caused by the sedentary lifestyle of both patients. The slight improvement in the quality of life perceived by E. could be influenced by the patient's clinical comorbidity of mood disorder.

Conclusion

This report aims to contribute to the need to develop uniform exercise guidelines for people with CMT, highlighting the clinical benefits and improvements in performance in patients performing tailored exercises.

Conflicts of interest: The authors certify that there is no conflict of interest with any financial organization regarding the material discussed in the manuscript.

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Institutional Review Board: The research was approved by the head of the Neurology dept. of the local hospital in Genova, (Italy) and approved by the local bioethics committee.

Informed consent: It was obtained from all subjects involved in the study. Written informed consent has been obtained from the patients to publish this paper.

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