

CASE STUDY ON THE ROLE OF NEUROMOTOR RE-EDUCATION METHODS IN THE TREATMENT OF CHARCOT MARIE-TOOTH DISEASE

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Abstract: *The isolated Charcot disease occurs in young people and has a motor onset, being a touch of motor cells, anywhere in the cerebrospinal axis. The paper is a case study of a subject diagnosed with Charcot Marie-Tooth disease. The physical therapy intervention aimed to apply elements of neuromotor re-education methods over a period of nine months. The results reveal that combining neuromotor re-education methods contributes to improving muscle elasticity and capsuloligamentous structures, thus preventing the formation of dysfunctional positions, and induces voluntary motor activity in the affected muscles, thus improving muscle imbalances specific to pathology.*

Key words: *methods, re-education, motor activity, imbalance.*

1. Introduction

Charcot Marie Tooth is a heterogeneous group of genetic disorders, being the most common hereditary peripheral neuropathies in the world, with approximately 10 to 28 cases for every 100,000 newborns [2]. The condition presents several genetic subtypes and clinical aspects: type 1 (CMT1), type 2 (CMT2), type 3 (CMT3), type 4 (CMT4), type 5 (CMT5), type 6 (CMT6), X-linked (CMTX) and intermediate [8]. Dominant and recessive demyelinating forms are classified as CMT type 2 (CMT2) and autosomal recessive forms are classified as CMT 2 (AR-CMT2) [7]. Although patients usually have the first symptoms

between the first and second decade of life, several other clinical manifestations may appear later. In addition, clinical severity can be variable, fluctuating between an extremely mild form of the disease that may remain unknown, to a more severe form associated with weakness and disability [8]. The typical symptom present is hypotonia in the legs, and evaluation may reveal tendon reflexes absent on the dorsal flexion movement. The damage may be bilateral with symmetrical atrophy of the calf muscles. Lifespan is not reduced [10], [3]. In most studies, the treatment approach is symptomatic. Affected individuals are

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evaluated and the intervention is multidisciplinary, including specializations such as: neurology, orthopaedics, surgery, physiotherapy, occupational therapy, etc. [1], [4], [6].

2. Goals

The main goal was to prove the effectiveness of neuromotor re-education methods in relieving symptoms in a patient diagnosed with Charcot-Marie-Tooth. Other goals were:

Selection of re-education methods to improve the symptoms of the disease in question;

Ordering them in a logical sequence within kinesitherapy programs, so as to achieve rehabilitation with maximum efficiency;

Identifying optimal ways to apply selected and customized methods improve functional capacities.

3. Material and Methods

When developing this paper, we started from the idea that by applying re-education methods, following the stages of motor control, intervention can be made in order to improve symptoms.

Starting from this premise, we proposed the following hypothesis:

It is assumed that the application of neuromotor re-education methods, following the stages of motor control, contributes to maintaining functional capacities and reducing symptoms in Charcot-Marie-Tooth disease, triggering beneficial effects on the entire body.

The research was conducted on a single female patient diagnosed with Charcot-Marie-Tooth disease. Below we will present the detailed individual file,

consisting of disease history, clinical examination, laboratory investigations, evolution and medical recommendations:

Information data of the subject

Initials: S.E.

Age: 27 years

Clinical diagnosis: peroneal muscular atrophy, Charcot-Marie-Tooth disease

Personal history: had muscle weakness from the first years of life;

Family history: Family history does not reveal other cases of pathology associated with that of the patient;

Mechanism of occurrence: autosomal dominant genetic disease;

Drug treatment: does not undergo;

Physiological parameters: within normal limits;

Medical recommendations: avoid overexertion.

Anamnesis: The 27-year-old patient with bilateral neurological crooked leg, sequelae of Charcot-Marie-Tooth disease, was operated on the right side 8 months ago, and, in 2022, she completed specialized orthopaedic treatment for the left leg - Achilles tendon elongation. Postoperative evaluation was good, and immobilization was performed in gambieropodal circular cast.

The kinetotherapeutic intervention was performed over 6 months, between April and September 2023. The measurement methods consisted of: Attitude test, Static and dynamic balance test: *Romberg test*, *Measuring the support base in orthostatism*, *Berg balance scale*, Coordination testing for upper limbs: *pinwheels test*, *Index-nose test*, and for lower limbs: *Heel-knee test*, *Test of drawing the figure eight on the ground*, *Gait examination: Tinetti gait test.*

Functional diagnosis: the patient

presents atrophy at the level of flexor muscles of hands and legs and hypotonia at the level of arms, forearms and calves, antalgic reflex position of flexion of upper limbs, deficient stability, coordination and ability, walking is unsafe and performed with a wide support base.

The treatment stages followed the stages of motor control, as follows:

Stage I aimed to improve the amplitude of motion and prevent dysfunctional positions;

In *stage II* we aimed to improve overall muscle strength and, analytically, in the lower limbs; [5]

In *the third stage* we achieved the improvement of proprioception and balance;

The fourth stage was represented by the improvement of controlled mobility, aiming for the patient's evolution to consist in the ability to execute movements during any posture, to load by body weight with fixed distal segments or to rotate the head and torso around the longitudinal axis during these postures. This is also the stage in which we pursued the re-education of walking;

The fifth stage, representing the last level of motor control - the highest, aimed to improve ability, focusing on the re-education of fine motor movements in both the upper and lower limbs.

The goals of the kinetotherapeutic intervention were as follows:

- Maintaining kinaesthetic image;
- Maintaining joint mobility and preventing dysfunctional positions;
- Induction of voluntary motor activity in the affected muscles;
- Improving muscle strength;
- Improving stability, coordination and controlled mobility;

- Improving ability by bilateral training of upper and lower limbs;
- Re-education of gait with reduction of ataxia.

Kinetherapeutic intervention:

At the beginning of each treatment session, we aimed to relax the muscles by applying stretching techniques combined with facilitation techniques [9] such as rhythmic initiation (IR) and rhythmic rotation (RR). We have analytically worked each joint, starting with the proximal one and each shortened muscle group.

With *the Bobath method*, at the level of the *upper limb*, we followed the stretching of the muscles by maintaining inhibitory reflex postures, from the patient's position of dorsal decubitus: external rotation of the shoulder, supination of the forearm with elbow extension; horizontal abduction, external rotation of the shoulder and supination of the forearm with elbow extension; abduction of the upper limb over horizontal with external rotation; extension of the shoulder with external rotation in diagonally in the posterior plane, from the patient's position of lateral and seated decubitus; abduction of the thumb finger with external rotation of the shoulder, supination of the forearm and extension of the elbow. For the *lower limb* we performed the following: flexion, abduction, external rotation of the hip and dorsal flexion of the leg, from dorsal decubitus; external rotation of the hip with the knee extended from the dorsal decubitus; dorsal flexion of the fingers (4 external fingers); We passively kept these reflex inhibitory postures until the onset of pain, and gradually increased the amplitude.

In the *lower limbs*, the techniques used were rhythmic initiation (IR) and rhythmic rotations (RR) in the axis of the segment, targeting the entire limb, and analytically, processing each joint (hip, knee, ankle).

In *stage II* we aimed to improve overall muscle strength by inducing voluntary motor activity in the affected muscles and maintaining the kinaesthetic image [11]. In order to achieve the goals of this stage of treatment, we applied as means, facilitation techniques on Kabat diagonals for both upper and lower limbs. In the upper limbs, we used the diagonal D1 of extension and D2 of flexion. Thus, on the diagonals D1 of extension and D2 of flexion at the level of the upper limbs, we started with the adapted rhythmic initiation (IR) technique (passive movement - passive-active - active), gradually moving to the sequential techniques for strengthening (SC), agonistic reversal (IA), and isometric contraction in the shortened area (CIS) at the end of the amplitudes of motion. The diagonals were made with both "shoulder pivot" and "elbow pivot". In doing so, we constantly requested eye contact and patient attention to the movements performed.

At the level of the lower limbs we followed the same principles, applying the diagonals D1 and D2 of flexion. Since these movements require great muscular strength, we first applied using the knee as a pivot, fragmenting the movement halfway through the stroke, thus aiming to tone the quadriceps. Taking the same path as in the upper limbs, we applied to these diagonals the following facilitation techniques:

- repeated contractions (CR) on D1F and D2F with "knee pivot", for quadriceps, and the return movement was achieved by the

agonistic reversal technique (AI), which uses eccentric contraction, following a step-by-step pattern, thus toning the muscles in conditions of elongation and shortening in a single movement.

- on the aforementioned diagonals, but with "hip pivot", we applied the sequencing for strengthening (SS) for the quadriceps and dorsal flexors of the foot.

- for faster toning, at the ends of the motion amplitudes we used isometric contraction in the shortened area (ICS) with variable holding of 4-6 seconds.

To improve *coordination*, we also used from the Kabat method the *bilateral symmetrical diagonals* for the upper limbs and lower limbs in the extension direction (outside the segment).

In order to increase the force at the *trunk* level, we aimed to promote the nerve influx from the strong muscles of the upper trunk to the weak muscles of the lower trunk, using the techniques of "chop" or "lifting" on the Kabat diagonals, which we made from dorsal decubitus. With one distal grip on the hands and another on the forehead and neck, we resisted while performing the slow reversal (SR) and slow reversal with resistance (SRR) techniques. They have been used to tone abdominal muscles and lower trunk extensors. We achieved increasing the force in the upper limbs at the level of the descending and adductor muscles of the shoulders (great dorsal, great round, great toothed, great pectoral), as well as brachial triceps, from the patient's position of elongated sitting, with support on the palms and grips at the pelvis level, applying the facilitation techniques of slow reversal (SR), slow reversal with resistance (SRR), alternate isometry (AIs) and isometric contraction in the shortened area (ICS).

In the *third stage* we achieved the *improvement of stability, balance and coordination* using specific facilitation techniques on Kabat diagonals, and then, from several positions such as: elongated sitting, shortened sitting, quadruped, kneeling, and orthostatism.

We used all diagonals (D1F, D1E, D2F, D2E) on both upper and lower limbs. First, we asked the patient to perform active mobilization, then we applied progressive resistance, and during the arc of motion, we applied the techniques of facilitating alternate isometry (AIs) and rhythmic stabilization (RS), initially using a single technique, and then, for complexity, we used both, thus stimulating the patient's attention, both to movements and verbal commands (for example: "pull", "don't let me move your arm", "pull", "hold", "push", etc.).

To improve trunk stability *from the elongated sitting position*, hand support was maintained at the back of the body. The posterior positioning of the arms increased the support base, making it easier for the patient to maintain. To relieve it, I asked the subject to flex the head a lot, thus the centre of gravity falling inside the support base. From this position, the patient actively transferred weight laterally left-right, then front and back, as well as in an oblique right-front, left-back position. Subsequently, we applied manual resistance, performing the facilitation techniques in the following order: slow reversal (SR), slow reversal with resistance (SRR), isometric contraction in the shortened area (ICS), alternate isometry (AIs) and rhythmic stabilization (RS). We used the same model from the patient's *shortened seating* position to diminish the support surface.

From the position of the *quadrupedal* patient, we performed left-right, anterior-posterior and oblique loading, first actively, then by applying the techniques of facilitating slow reversal (SR), slow reversal with resistance (SRR), alternating isometry (AIs) and rhythmic stabilization (Rs) on the aforementioned directions. From the same position I asked the subject to perform the flexion of an upper limb, and then, of a lower limb, as to maintain the position with three points of support, in order to achieve progressivity. Then, from the quadrupedal gait, we applied the technique of progression with resistance (PR) back and forth, holding the grips both at pelvis and shoulder level.

To increase progressivity, from the *"knee" position* we used the same techniques as in quadrupeds (SR, SRR, AIs, RS) with the grips held at the pelvis level, but preceded by active mobilization.

With the *Margaret Rood method*, we used pressure to increase proprioception. These were made with the aim of stimulating articular proprioceptors. The presses were made with a force along the axis of the extremities, both in the open kinematic chain from the dorsal decubitus, and in the closed kinematic chain from the quadrupedal position, as follows: pressure on the shoulders from the sitting position, pressures on the scapular and pelvic girdles from the quadrupedal position.

The fourth stage was represented by the improvement of *controlled mobility*. To re-educate walking we applied elements from the *Frenkel method*, as follows:

Seated exercises:

1. Touch with your foot (toe or heel) various points marked on the floor or indicated.
2. Getting up from a chair and sitting back.

- at command 1 gently flex the calves below the edge of the seat;
- at command 2 gently bend the head and torso forward;
- at command 3, rise, with extension of the knees and hips.

Balance exercises

1. Standing with the upper limbs extended, maintain the position with a book on the head for as long as possible.
2. Maintaining unipodal balance for 3 seconds.
3. From orthostatism, with the base of support smaller and smaller, and with the arms beside the body, move the head up and down and from right to left.

Exercises to re-educate walking

In the position of standing. In this position, we did the re-education of walking on diagrams drawn on the floor. Re-education of walking begins with lateral walking, considered easier because it can be helped by body balance, and then walking in a straight line, the patient stepping on coloured soles. The return was made according to a diagram in the shape of a circle, drawn on the floor. The patient learned to turn by moving leg after leg, a quarter of the entire rotation so that she could perform a 180° turn in 2 moves (2 steps).

Coordination exercises

1. Visual motor coordination. Left-right progression, essential for reading, without moving the head while a ball moves on a table from left to right, the patient should try to focus the moving object.

2. Standing in front of a mirror, the patient alternately raised her right hand and left leg and vice versa.
3. From dorsal decubitus, the patient raised the left leg and right hand vertically, then repeated for the right leg and left hand.
4. Standing face to face with the patient, with a small ball, we helped her transfer it from one hand to another. The movement was progressively performed in front or up above her head and repeated until she was able to execute the movement correctly.

We also applied the technique to facilitate progression with resistance (PR) with grips at pelvis and shoulder level while walking back and forth.

The fifth stage represents the last level of motor control - the highest, aimed to improve ability, focusing on the re-education of fine motor movements in both the upper and lower limbs.

In this regard, we aimed to re-educate fine movements of the upper limbs, finger gripping and clamping, and on lower limbs the goal was to achieve correct gait, stepping over obstacles, etc. Thus, we applied the normal sequential technique (NS), directing various fine movements with a slight manual resistance, having the role of guiding the motor action.

4. Results

In this subchapter, the results obtained were analysed, and the values recorded in the tables highlighted a series of aspects regarding the efficiency of neuromotor re-education methods.

Static balance test

Table 1

Test applied	Static equilibrium testing	
	Initial testing	Final testing
Romberg Test	In this test I wanted to find out what the patient's balance status was, so in the initial test I positioned the patient standing with her legs close together and asked her to close her eyes. The patient managed to hold the position for 15 seconds, subsequently losing her balance. I tried to apply the second version of the test, the patient being also positioned in orthostatism, but now the legs were positioned in front of each other. This time, she only managed to hold the position for 12 seconds after I told her to close her eyes, then she lost her balance.	At the end of the recovery period, I reapplied this test to check if the static balance had improved. Thus, we tried both versions of the test, the patient being also positioned in orthostatism. With her legs close together and her eyes closed, this time she managed to maintain the balance position for about 30 seconds without swaying, then made a few oscillations and subsequently lost her balance. As for the second version, the patient managed to hold the position for 20 seconds, then took a lateral step with the foot positioned in front so as not to lose her static balance.

• Measuring the Support Base in Standing position

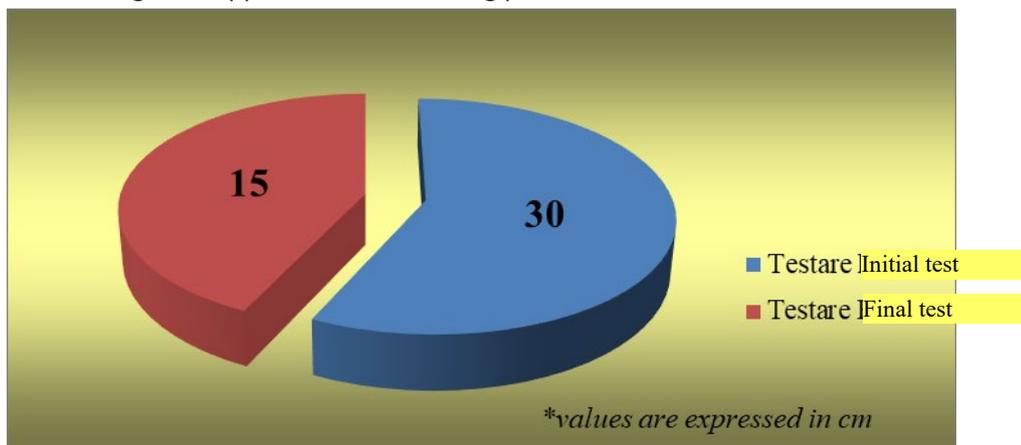


Fig. 1 Measuring the support base

Measuring the support base is very important in the kinetherapeutic

program, due to with the help of this measurement we manage to realize the patient's self-confidence. In Fig. 1 we see

that at the initial test the distance between the heels of the feet was 30 cm, which represents a neurological imbalance that affects both muscle groups and the patient's mental state. Subsequently, at

the final test, the standing support base was reduced to 15 cm, which represents progress following the proper application of the treatment.

- *Berg balance scale*

Berg scale results

Table 2

Action	"Berg" balance scale	
	Initial testing	Final testing
Seated without support	4	4
Lifted from seated	2	4
Standing without support	2	4
Standing with eyes open	4	4
Standing with legs close together	1	2
Stretching forward (hand at 90 degrees, number of centimetres)	1	3
Lifting an object off the ground	1	3
Turning to look over your left and right shoulder	1	3
360-degree turn	1	2
Alternating seat touch	2	3
The position in which one leg performs the movement	1	2
Standing on one leg	0	0
Sitting	3	4
Transfer	3	4
Total	26	42

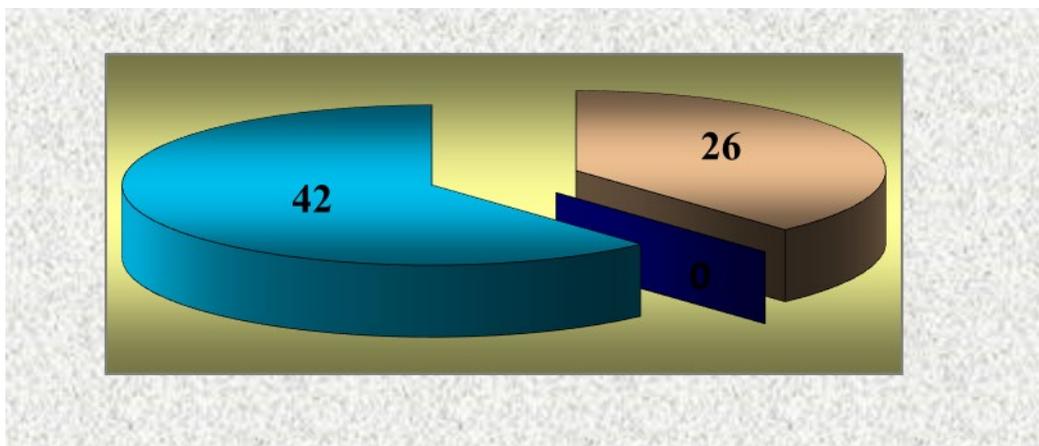


Fig. 2. *Dynamics of balance evolution – Berg test*

Balance is a very important component used in all daily activities of a person. That's why we tested patient balance using the Berg test. This pointed out that in the initial testing the subject was able to perform certain actions with difficulty. She was able to stand upright, but only after several attempts, and then she held the position for 30 seconds without support. Shrinking the support base, she needs help to take the position, but without much success in maintaining it. Also, from orthostatism, she tried to reach over to pick up an object, but she constantly had

to be supervised, so as not to get unbalanced. As for turning 360°, she needs close supervision or verbal guidance. At the same time, she cannot maintain the standing position on one leg, but in terms of transfer and sitting she is able to handle controlling movements with her hands. After the recovery period, during which a kinetotherapeutic program adapted to the patient's situation was applied, we applied Berg's test once again, the results being better, the final score was 42 points, compared to the initial one of 26 points.

- Coordination testing

Upper limb coordination test results

Table 3

Test Applied	Upper limb coordination testing	
	Initial testing	Final testing
Pinwheel test	She managed to do the test for a short time, 3-5 seconds, then lost pace.	She managed to do the test much easier, the pace of execution was better, and the time increased to almost a minute.
Index-nose test	She keeps the arm in abduction, but when she wants to take it to the front to perform the test, oscillation movements occur, the trajectory not being a precise one.	She keeps the arm in abduction, and when performing the test, the trajectory of the arm is no longer oscillating, managing to touch the nose with the index.
Puppet Challenge	The patient managed to perform these movements for a short time, 10-15 seconds, later saying that she was tired.	The patient now manages to do the test, the movements being more defined and the execution time has increased to one minute.

Lower limb coordination test results

Table 4

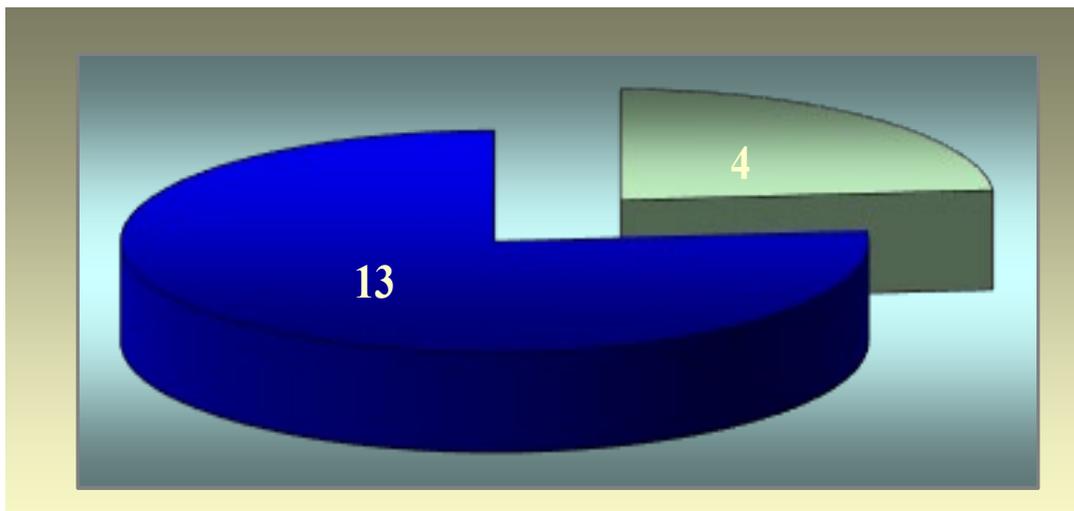
Test Applied	Lower limb coordination testing	
	Initial testing	Final testing
Heel-knee test	The patient managed to perform this test only for a few 4-5 attempts, later saying that it is hard and that she tires.	The patient was able to perform the test, the movements being more accurate, and the execution time increased to about 50 seconds.
Test of drawing the figure 8 on the ground	The patient was only able to draw half of the figure 8 because she was out of balance.	The patient managed to draw the number 8 entirely, performing the test several times, without losing her balance.

- Gait examination

Gait examination results

Table 5

Action	Tinetti gait test	
	Initial testing	Final testing
Balance in sitting	0	1
Getting up	1	2
Trying to get up	1	2
Balance immediately after getting up (first 5 seconds)	0	1
Balance in sitting	1	2
Pushing (subject in maximum position with legs as close together as possible; the examiner holds palms or hands on the subject's chest, and it gently pushes the subject 3 times)	0	1
Standing with her eyes closed	0	1
Turning 360 degrees	0	1
Sitting	1	2
Total	4	13

Fig. 3. *Dynamics of gait evolution*

5. Discussion

Walking is the most precious thing, and many people would do anything to walk again. That's why I did my best to help this patient regain her functional capacity. The gait examination involved two tests, one initial and one final. During the initial test, we found that the balance in sitting was deficient, and getting up was done with the help of the arms, needing several attempts. After she managed to get up,

she maintained the standing position with difficulty, because there were oscillations in the trunk. In the end, with help she managed to maintain the position, but with a large area. After stabilizing the position, I asked her to close her eyes, she did it and immediately began to wobble, no longer having stability. When sitting, she used her arms. In the second test, namely the final one, the patient became

self-assured, managing to perform all the actions on her own. In terms of result, there was a final score of 13 points compared to the initial score of 4 points.

Starting from the attitude examination, the kinetherapeutic intervention followed had beneficial influences on the health of the subject, correcting some vicious postures both in the trunk and limbs. The treatment based on the application of the Kabat method, and facilitation techniques, following the stages of motor control, contributed to the improvement of movement amplitudes, stability and balance both static and dynamic, aspect proven by the results obtained at the "Romberg" evaluations. Due to the development of muscle strength, the patient can maintain the orthostatic position with a smaller support base (15 cm), compared to 30 cm initially. With regard to the results of the "Berg" rating scale, there is also an improvement in balance and coordination due to the application of treatment from different positions and with progressive loading. The deficiency that the patient has at this moment is the maintenance of unipodal support, but it does not affect her very much in daily life. Coordination also improved, meaning that, due to the application of analytical techniques, but also in the overall scheme, on diagonals, several muscles were involved in movement, which helped induce voluntary motor activity also in the affected muscles, so that the selected tests were executed at a faster pace and with more organized movements both in the upper and lower limbs, at the end of the intervention.

The analysis of the gait components was carried out using the Tinetti gait test. Thus, during the initial testing, we found balance problems in walking. The analysis of these components specific to the Tinetti test was performed at the usual

execution speed of the patient, not trying to increase the pace or speed of achieving the items. The loss of balance in standing and walking, the problems in getting up and sitting on the chair, the impossibility of performing various trivial actions have seen a considerable improvement due to the observance of the principles of re-education, giving the patient for the first time, the imprinting in the motor engram of the gesture of stepping, through repetition, then respecting progressivity, we switched to lateral walking in three strokes to facilitate the subject's awareness of the complex action of stepping. We only started to re-educate the back-and-forth gait when the laterality movements were no longer disorganized, which led to long-term maintained positive results.

4. Conclusions

Following the research and the results achieved, it can be stated that the initially established hypothesis has been confirmed. Due to this aspect, the conclusions of this paper are not intended to be generally valid, but are the conclusions drawn from the research carried out:

The hypothesis formulated above has been confirmed, meaning that combining neuromotor re-education methods contributes to improving muscle elasticity and capsule ligament structures, thus preventing the formation of dysfunctional positions. Carefully selected and appropriately adapted facilitation techniques induce voluntary motor activity in the affected muscles, maintain the kinaesthetic image and lead to motor response.

The application of inhibitory positions of the Bobath method and rhythmic initiation and rhythmic rotation techniques encouraged the reduction of hypertonia and the improvement of mobility by increasing

the amplitudes of movement at the level of the joints of the affected hemibody.

Muscle tonicity and trophicity was increased by applying the Kabat method in combination with facilitation techniques, thus improving muscle imbalances specific to pathology. The elements of the Margaret Rood method and the specific facilitation techniques applied in the open and closed kinematic chain, stimulated the articular proprioceptors, contributing to the re-education of stability, balance and coordination.

The re-education of gait was performed with elements of the Frenkel method, promoting the achievement of controlled skills with greater functionality.

The kinetherapeutic intervention also had beneficial effects on the patient's psyche, as she noticed from the first sessions that muscle strength and stability in walking were recovered.

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