Longitudinal Outcomes in Charcot-Marie-Tooth Disease: A Case Study Examination of Progressive Symptoms and Interventions

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***Abstract*—Background: Charcot-Marie-Tooth disease (CMT) encompasses a collection of genetic disorders that impact the nerves linking the brain and spinal cord to other parts of the body. CMT is the most prevalent inherited neuropathy. CMT impacts the sensory and motor nerves in the arms, hands, legs, and feet. Sensory nerves transmit information to the brain regarding sensations such as touch, temperature, pain, or pressure. Motor nerves are responsible for regulating muscle movements. When motor nerves deteriorate, they become incapable of sending signals to the muscles. The case study emphasizes how progressive CMT is in controlling symptoms and enhancing quality of life; extensive therapeutic procedures such as strengthening, stretching, and functional training were used. The aim was to investigate the effectiveness of physiotherapy on a person with Charcot-Marie-Tooth disease. Case description and methods: A 27-year-old male with Charcot-Marie-Tooth disease since May 2024, complaining of pain, difficulty in walking, and shivering of legs after 1km. Physical examination, including ankle, knee, and hip muscle strength; sensory evaluation of foot and joint range of motion; self-reported assessment of pain, and muscle atrophy noted in bilateral lower extremities. Findings and outcomes: At the end of the program, lots of improvements are observed. Currently, he can maintain a good spinal curvature while sitting. As the strength of the bilateral lower extremities and hip girdle muscles was weak, he underwent the AAROM exercises for the bilateral lower extremities. Conclusion: Patients with Charcot-Marie-Tooth illness must be managed in physical medicine and rehabilitated as soon as possible following a clinical evaluation of their joint, muscular, and sensory health to create a program tailored to each individual.**

***Index Terms*—Charcot-Marie-Tooth disease, Neuropathy, Physiotherapy, Strength training, Functional rehabilitation.**

I. INTRODUCTION

Charcot-Marie-Tooth disease Initially known as "peroneal muscular atrophy," Charcot-Marie-Tooth disease (CMT) was initially identified in 1886 by Tooth in London and Charcot and Marie in Paris.[1] Charcot-Marie-Tooth disease (CMT) encompasses a collection of genetic disorders that impact the nerves linking the brain and spinal cord to other parts of the body. CMT is the most prevalent inherited neuropathy. CMT impacts the sensory and motor nerves in the arms, hands, legs, and feet. Sensory nerves transmit information to the brain regarding sensations such as touch, temperature, pain, or pressure. Motor nerves are responsible for regulating muscle movements. When motor nerves deteriorate, they become incapable of sending signals to the muscles. As the most prevalent neurogenetic illness and most commonly inherited neuropathy, it is part of a clinically and genetically heterogeneous group of hereditary motor and sensory neuropathies, with an incidence of ½,500 people.[2]

The main clinical features of this disorder are usually slow progressive weakness, onset in childhood, familial occurrence, and muscular atrophy affecting the legs and feet.[3] Later on, the hands may also be affected, and further clinical features may include depression of tendon reflexes and mild to moderate distal sensory impairment.[4]

Hypotonia and hyposthenia of the leg and foot intrinsic muscles result from CMT, which affects both the upper and lower limbs and gradually spreads to the hands and forearms.[5] Owing to the deformed feet and altered

muscle balance, stance and/or swing phase function, as well as the capacity to heel and toe walk, have been used to characterize gait patterns, which are indicative of strength-related deficits.[6]

Walking difficulty, ankle twisting, tripping, postural instability, and frequent falls are common complaints from patients.[5,6] In addition, the illness can cause changes in sensory and respiratory functions as well as less common symptoms, including cramps, exhaustion, and discomfort that can have a serious impact on a patient's quality of life.[7]

II. CASE PRESENTATION

A 27-year-old male presented on 20th May 2024 with the chief complaint of difficulty walking, as his leg started shivering after 1km. Both legs have had pain from the foot to the knee for 2 years. He has been a known case of hereditary motor sensory neuropathy disease since May 2024. In their family paternal grandmother, along with the father and paternal aunt were affected clinically and symptomatically, while the mother is normal.

III. ASSESSMENT AND EVALUATION

On observation, there was a slight loss of lumbar lordosis Mild pes cavus foot was noted in the bilateral lower extremities in the high sitting position. During long sitting and supine lying the left hip was externally rotated, bilateral foot plantar flexed, with hallux abducted and flexed on the left side and abducted on the right side. There was symmetrical muscle wasting present over the bilateral extremities of the distal tibia part

On palpation, there was muscle atrophy noted in the bilateral lower extremities, left side -23cm (reference point 5cm from left malleolus) and on the right side was 21cm (reference point 5cm from right malleolus). He presented with normal muscle tone in the upper and lower extremities. The deep tendon reflexes were diminished in the right lower limb (knee and ankle jerk) and normal in the left side and the plantar response was extensor response type, with difficulty in eliciting. Normal reflexes in both upper limbs. Joint integrity and Muscle atrophy. He presented with crepitus in the bilateral knees occasionally, while in the ankle, it was constant. The range of motion he presented was associated with the hypermobility of the ankle joint. He has a full passive range but his active range of motion is limited in the ankle and subtalar joint due to muscle weakness.

Sensory examination revealed impaired sensation in the dermatomes L4, L5, and S1 for the temperature, whereas pinprick, superficial touch deep, and cortical sensation were intact muscle and endurance He presented with good muscle strength in the lower extremities. Distal muscular weakness was more than proximal muscles strength in lower limb .muscles Trunk flexor 5, trunk extensor trunk rotator 5 hip flexor left side 4/5, right side 4/5 hip extensor left4/5,right4/5 hip abductor left 4/5 right4/5 hip adductor left 4/5 right4/5 hip external rotator left 4/5 right 4/5 hip internal rotator left 4/5 right 4/5 knee flexor left 4/5 right 4/5 knee extensor left 4/5 right 4/5 dorsiflexor left 3/5 right 3+/5 plantar flexors left 3/5 right 3+/5 foot intrinsic muscle left 1/5 right 2/5 Trunk and pelvic stability. He presented with good strength of trunk muscles. He also has good trunk control while reaching forward, sideways, and backward to touch the target.

The pelvic stability was good. He presented with good static and dynamic sitting balance. He was able to assume standing independently but found difficulty in the narrow base of support after 15min _-20 min because of the onset of tingling and shakiness of bilateral lower extremities. He has difficulty in tandem stance and single limb stance gait analysis. He has normal step length and stride length with adequate cadence. He tends to have high-steppage Gait on the right side with hallux drop during the swing phase of the gait cycle. There is a more pronounced plantar flexed bilateral ankle joint during the swing phase.

IV. TREATMENT INTERVENTION

The goal of physiotherapy intervention is to improve the strength of lower limb muscles to improve balance and stability of ankle joints to improve the quality of gait educate the patient about pedal sensory loss and teach strategies to prevent injuries. Stretching exercises perform 5 reps with a hold of 20 secs Calf stretch, in a standing position stabilizes the knee joint with wall support. Now try to extend the other leg and keep the knee straight. Hamstring stretch in a supine lying position place one towel around the ankle keeping the other leg straight. Now try to pull the towards yourself. Plantar fascia stretch in a high sitting

position place your ankle other side of your thigh. Now hold the heel with one hand and with the other hand hold all the toes and pull towards your side. Hip flexor stretch place your hand in front of your foot or on your waist with your hip flexed and your knee 90 degrees and another leg extended backward.

Exercise to maintain balance and stability of ankle joint Tandem stance ball dribbling. In tandem stance drop the ball towards the ground and try to hold back the ball when it bounces back make sure to perform under assistance. 'Y' stepping put the colour mat patterns of Y as shown, and try to place the foot clockwise for the right leg and anticlockwise for the leg. Single leg cone touch In a standing position place one in front now touches the tip of the cone with an alternate great one. Lateral weight shifting makes him stand on the foam with the support and asks him to shift his body weight once left and once right side.

Strengthening of lower limb muscle Hip abductor Strengthening Assume the crook lying position over the bed and put the resistance bend around the knee. Try to open the leg against gravity and again come back to the original position. Hip extensor strengthening in a standing position with a 2kg weight cuff around the ankle joint lifts the leg backward. Gluteus muscle strengthening Assume the quadrupled position. Gently try to move the hip to the ceiling keeping the knee in 90-degree flexion. Hip abductor Strengthening place a pillow in between the distal end of your thighs now try to press it from both the legs.

Single leg bridges Lie down over the bed other back keeping one leg bent and the other leg straight. Try to lift the pelvis above the bed and hold the position for 5 secs. Lunges stand in spilled stance with the right foot roughly 2-3 feet in front of the leg foot. Your torso is straight your shoulders are back and down your one core is engaged and your hands are resting on your hips. Bend the knees and lower your body hold the position for 5 sec. Initially can take help for support and gradually reduce the support. For Squatting, walk your feet slightly forward your feet should be shoulder–width apart slide down until the thighs are parallel to the floor your knees should be over your ankles. Kneeling to standing in a full kneeling position and then half kneeling and then standing erect. Getting up from the lower surface sit down on the lower surface and try to get up without using the upper limb. Tibialis anterior strengthening in a high sitting position, extend your leg completely keeping the feet in neutral, and try to bring the feet closed towards your body.

Dorsiflexors strengthening in a long sitting position theraband wrapped around his forefoot trying to bring the foot closer towards your body. Simultaneously the other person will be pulling the theraband towards him. Evertor strengthening In a high sitting position place theraband around the foot firmly on the ground and now move the foot outwards. Intrinsic foot muscle place cloth underneath your foot and try to bring the cloth near to your body with a squeezing pattern movement from the toes. Hallux strengthening in a high sitting position feet well placed on the ground place two fingers on other toes and with another finger with active assistance extend the great toe. Toes play in a high sitting position with feet well placed on the ground. Try abducting all the toes of your foot hold for 10 seconds and then abduct it back. Exercise for postural Reinforcement perform a exercise 2 time a day and this position for 10 seconds with 10 repetitions. Dead bug exercise in a supine lying position brings your shoulder at 90 degrees and hip and knee at 90 degrees same side while engaging the core muscle. Bird exercise in a quadrupled position extend one arm of one side and extend the leg of the other side and then try to bring it near to touch the elbow and knee of the alternate side.

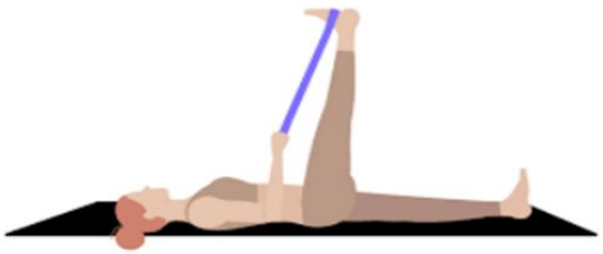


FIG 1- HAMSTRING STRETCH

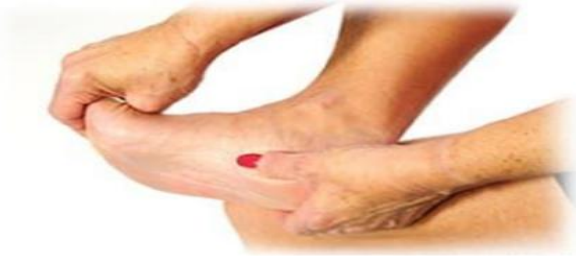


FIG 2 – PLANTAR FASCIA STRETCH

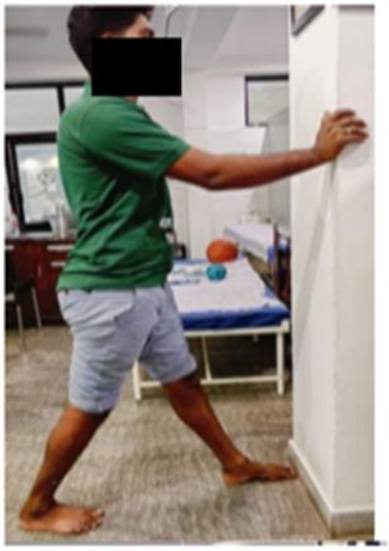



FIG 3 AND 4 - CALF STRETCH

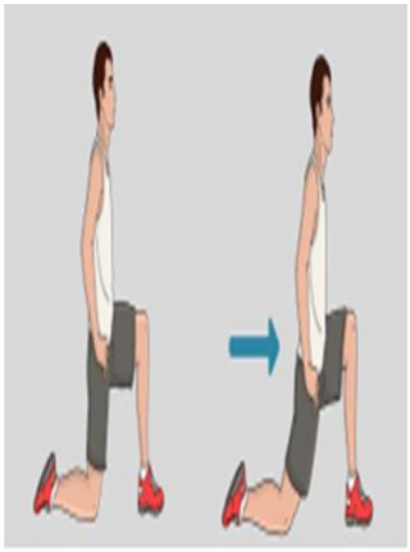


FIG 5-HIP FLEXOR STRETCH

FIG 6-TANDEM BALL DRIBBLING


FIG 7 – SINGLE LEG CONE TOUCH

FIG 8 –‘ Y’ STEPPING

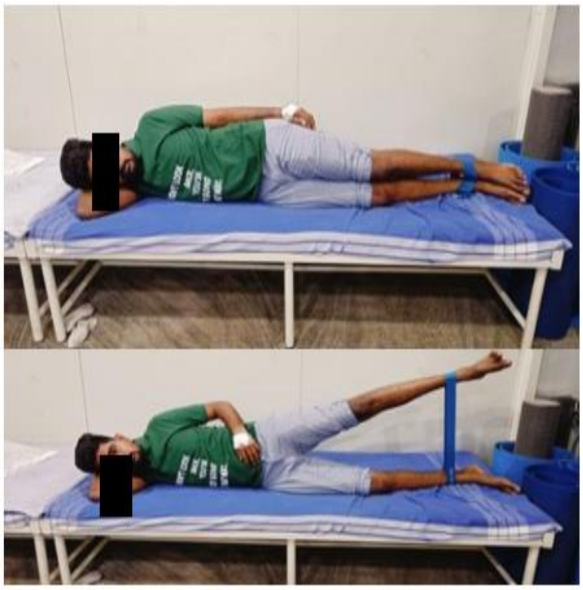


FIG 9-HIP ABDUCTOR STRENGTHENING

FIG 10- LATERAL WEIGHT SHIFTING

FIG 11-GLUTEUS MUSCLE STRENTHENING

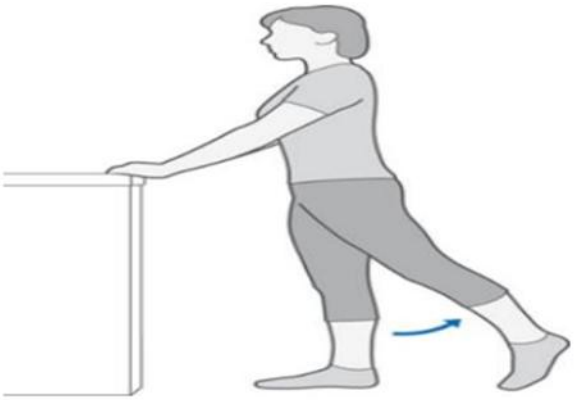


FIG12-HIP EXTENSORS STRENTHEN

FIG 13-GETTING UP FROM THE LOWER SURFACE

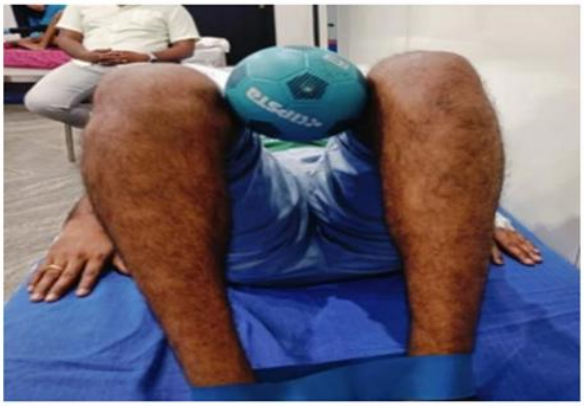


FIG 14 - HIP ADDUCTOR STRENTHENING

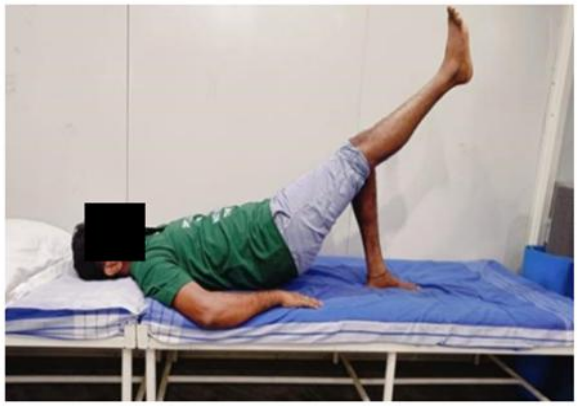


FIG 15- SINGLE LEG BRIDGING

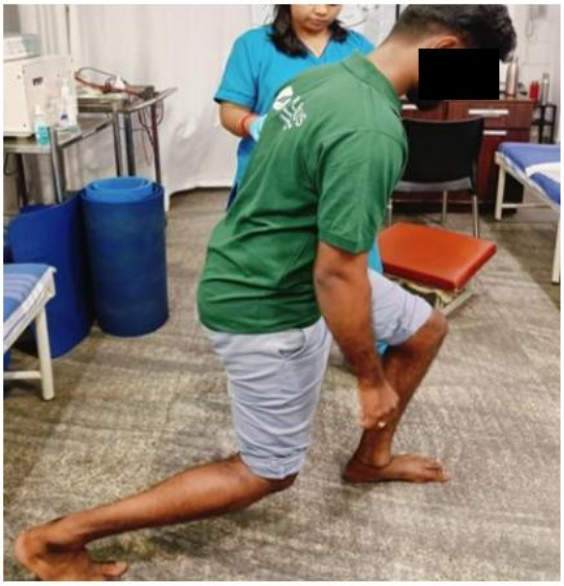


FIG 16- LUNGES

FIG 17- SQUATS

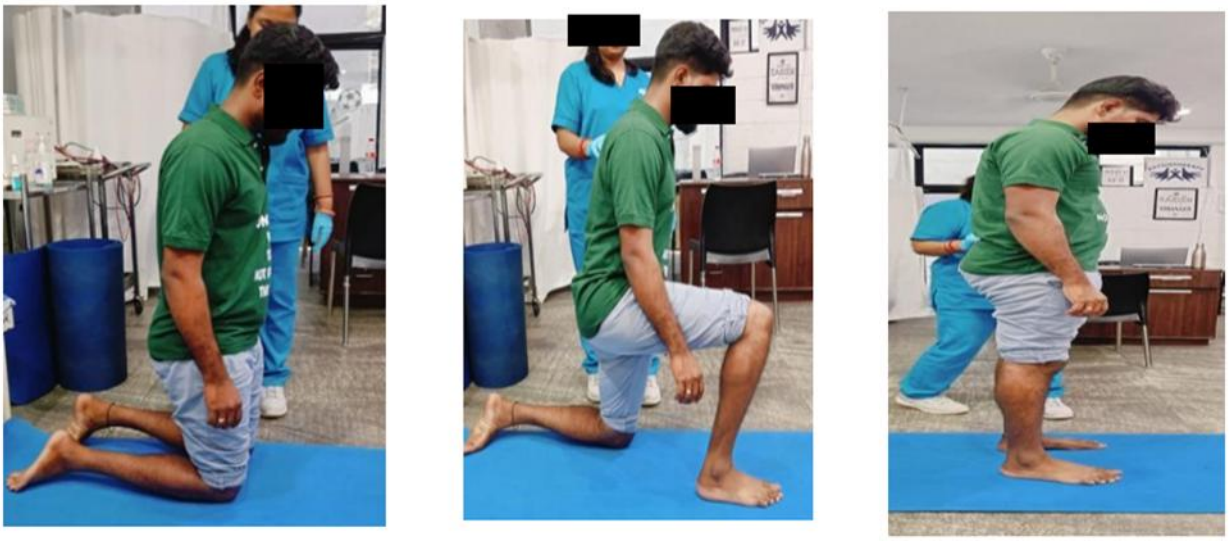


FIG 18- KNEELING TO STANDING

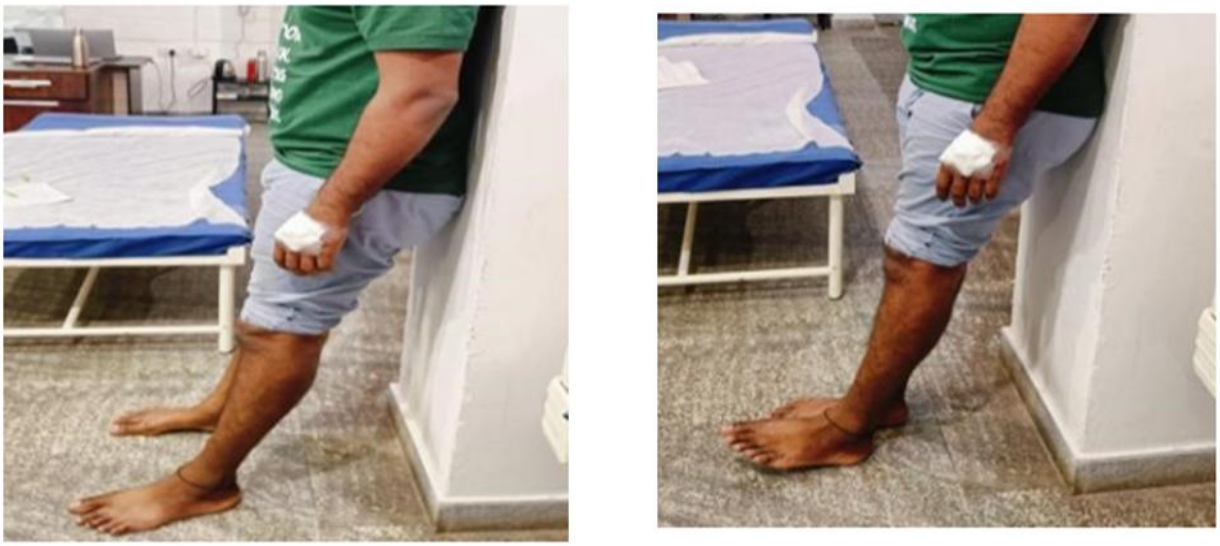


FIG 19-TIBIALIS ANTERIOR STRENGTHENING

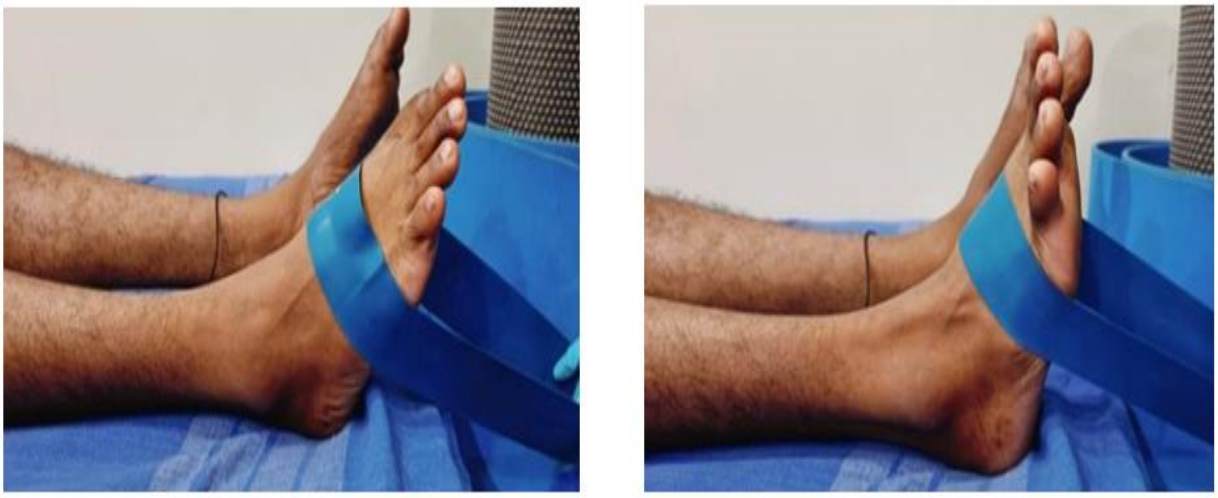


FIG 20-DORSIFLEXORS STRENGTHENING

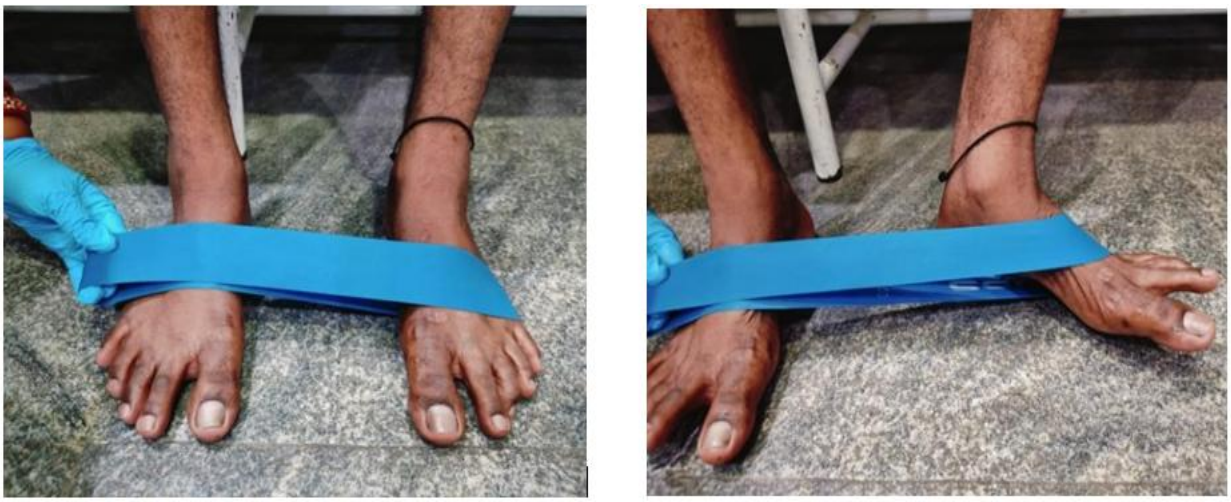


FIG 21- EVERTORS STRENGTHENING

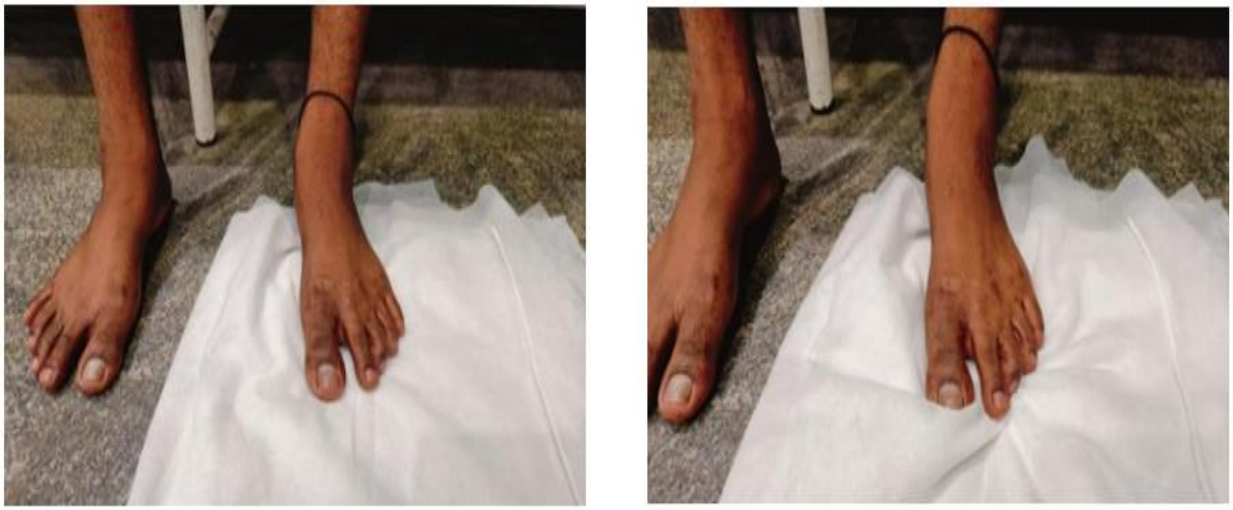
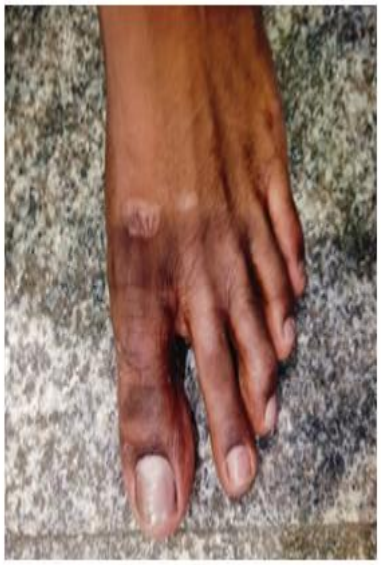


FIG 22-INTRINSIC FOOT MUSCLES

FIG 23-HALLUX STRENGTHENING



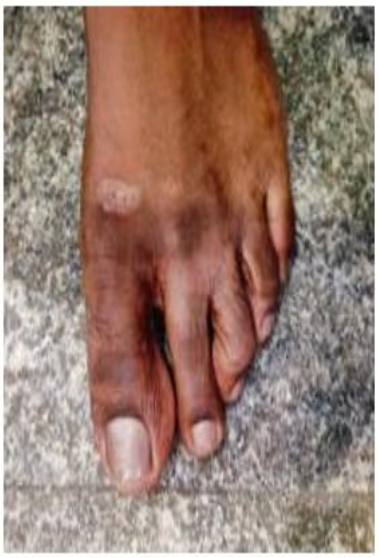


FIG 24-TOES PLAY


FIG 25-DEAD BUG EXERCISE

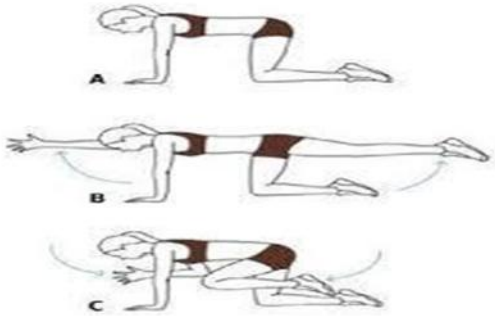


FIG 26-BIRD DOG EXERCISE

V. RESULT

At the end of the program, significant improvements are observed. Currently, he can maintain a good spinal curvature while sitting. As the strength of the bilateral lower extremities and hip girdle muscles was weak, he underwent the AAROM exercises for the bilateral lower extremities. Currently, the assistance while performing the hip AAROM has been reduced

He initiated performing the hip flexion-extension in the quadripod position without assistance. The strength and endurance of trunk muscles (trunk flexors, trunk extensors, trunk side flexors, and trunk rotators) have improved.

The improvements in trunk muscle strength lead to better trunk control while sitting, reaching out, and performing the exercises over the tilt table. Currently, he can hold the bilateral upper extremities overhead with good trunk control.

Overall, the dynamic sitting balance has improved. He underwent strengthening exercises for the shoulder girdle muscles. Improvement in trunk and shoulder muscle strength helped him to ease the transfer from the different surfaces. On the tilt table, he has initiated standing with the 90-degree inclination for 4-5 seconds without any assistance.

VI. DISCUSSION

Despite being an incurable condition, CMT patients' quality of life can be enhanced by symptomatic therapies such as physical therapy, surgery where necessary, analgesics, and various pain management techniques, among others.[8]

Patients with Charcot-Marie-Tooth illness must be managed in physical medicine and rehabilitated as soon as possible following a clinical evaluation of their joint, muscular, and sensory health to create a program tailored to each individual.[9]

Some research has shown that a medical and technical meeting, including a physical and rehabilitation physician, as well as a specialized shoe technician, is held to manufacture the orthopaedic shoes. They establish the specifications, including therapeutic goals, descriptions of the upper and plantar orthoses, the sole, the method of fastening, and other details for patients with Charcot-Marie-Tooth disease.

Some case studies demonstrated the value of consistent and successful physical therapy regimen in promoting functional independence in patients with Charcot-Marie-Tooth illness. There is evidence supporting the improvement in overall functional independence and the timing of ADL execution.

According to some recent research, bracing with OS may be a useful treatment for gait impairments in people with CMT.[10] In some cases, it has been observed that using various items in different forms allows for more functional tasks to be completed through gripping exercises. After achieving these goals and stretching tight muscles, the patient was eager to begin gait training, initially on a parallel bar

and then off of it. The plantar flexed assist was utilized. An orthosis is recommended to strengthen his dorsiflexor muscles and enhance his gait. After the patient's gait significantly improved, static cycling was used for endurance training.

Although surgery is typically necessary to treat foot abnormalities, clinical studies following the most common surgical method—triple arthrodesis—have demonstrated that the procedure has poor long-term outcomes in terms of pain alleviation and functional improvement.[11] It was discovered that a 24-week strength training program increased participants' walking speeds.[12]

Neither night splinting nor holding the ankle in the maximum dorsiflexion position for 6weeks resulted in any significant improvement in muscle strength or the passive range of movement around the ankle.[13]

In this study, we have tried to improve patients' ADL activities. Stretching, strengthening, aerobic exercises, and respiratory management were used in a comprehensive physiotherapy intervention for a CMT patient. The goals of the intervention were to preserve functional independence, prevent contractures, and improve quality of life. The patient initially displayed gains in muscle strength and range of motion, but as the illness worsened, these interventions became less successful, and the emphasis turned to controlling pain, averting complications, and optimizing involvement in everyday activities. To promote independence, the use of assistive technology and contextual adjustments became essential, underscoring.

VII. CONCLUSION

The study emphasises controlling symptoms and enhancing the quality of life for the person suffering from CMT; extensive therapeutic procedures such as strengthening, stretching, and functional training were used.

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