

The Use of the Richie Brace Dynamic Assist in a Patient with Charcot-Marie-Tooth Disease - A Case Report

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Abstract

Charcot-Marie-Tooth (CMT) disease, also known as hereditary motor and sensory neuropathy involves a group of diseases that effects the peripheral nervous system causing a range of symptoms such as foot drop and foot deformities. Management of CMT disease is based around the use Ankle Foot Orthosis (AFO) however, little research has been based around pes planus deformity and specific AFO's in adults as seen in our case. This case describes the use of the Riche Brace Dynamic Assist in a 36 year old female patient with CMT disease and pes planus deformity, assessing whether foot pain and functionality improved utilising the Foot Function Index (FFI) to assess pain and function after 3 months.

Keywords: Richie Brace Dynamic Assist; Charcot-Marie-Tooth Disease; Case Report

Introduction

Charcot-Marie-Tooth (CMT) disease is a group of inherited neuropathic diseases that primarily affects the peripheral motor and sensory nerves (1). This leads to progressive nerve damage, particularly in the lower limbs, often resulting in muscle weakness and severe foot deformities. Intrinsic muscle atrophy arises due to peripheral neuropathy which contributes to flexion deformities such as hammer toe and pes cavus (2,3). However, a proportion of individuals present with pes planus, usually diagnosed with CMT-2, resulting from anterior, peroneal and intrinsic muscle atrophy, secondary to degeneration of the common peroneal nerve as presented in this case (4).

Treatment for CMT is centred on rehabilitation and symptom management as there are currently no disease modifying therapies that can alter its future progression (5). Ankle foot orthosis (AFO) have been described in the management of individuals with CMT disease, in particular one that maintains dorsiflexion to aid those with severe muscle weakness and foot drop (6). The Richie Brace Dynamic Assist is a custom moulded hinged AFO which facilitates controlled dorsiflexion of the ankle (6,7). It is advised for use in individuals with foot drop, as seen in CMT, but there is paucity of data for its use in patients with this condition (7).

This is a case study of a patient with CMT disease, assessing the impact of the Richie Brace Dynamic Assist on pain severity and function utilising the Foot Function Index (FFI).

Case Presentation

The patient is a 36 year old female with a family history of CMT disease. Her mother has the condition. She had been experiencing constant severe pain on the sole of her foot with a considerable build up of callous causing her discomfort.

On physical examination there was weakness of the peronei with reduced ankle reflexes further highlighting peripheral nerve degeneration. There was calf inflexibility with reduced ankle dorsiflexion. On standing, there was a low arch profile as seen with a pes planus type foot deformity with an 'inverted champagne bottle' appearance from the calf to ankle (*Figure 1*) (8).

On visual gait analysis, there was a foot drop with inversion through swing, resulting in a high step and to allow the foot to clear the ground. As a result, there was a forefoot strike, followed by heel strike. During late swing / early stance there was hyperextension of the hallux with increased pronation in latter stance (*Figure 2*).

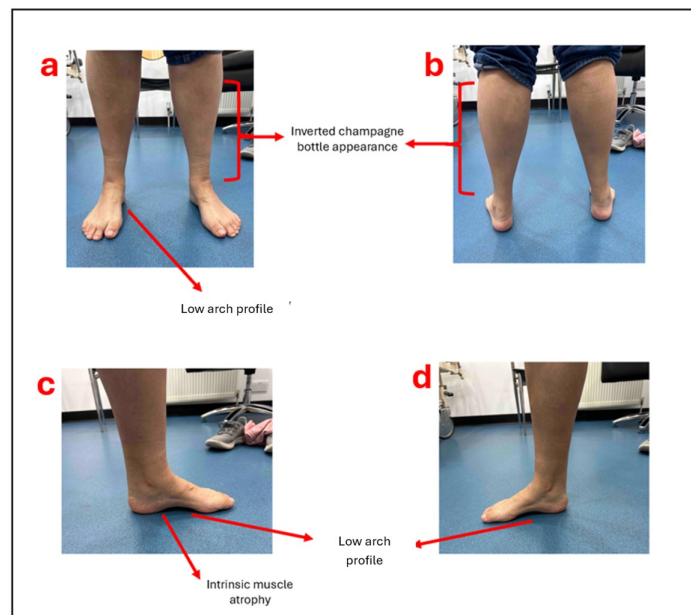


Figure 1- (a-d) Views of the patient with annotated signs indicative of Charcot-Marie-Tooth (CMT) disease **a)** anterior view **b)** posterior view **c)** Left foot, medial view **d)** Right foot, medial view.

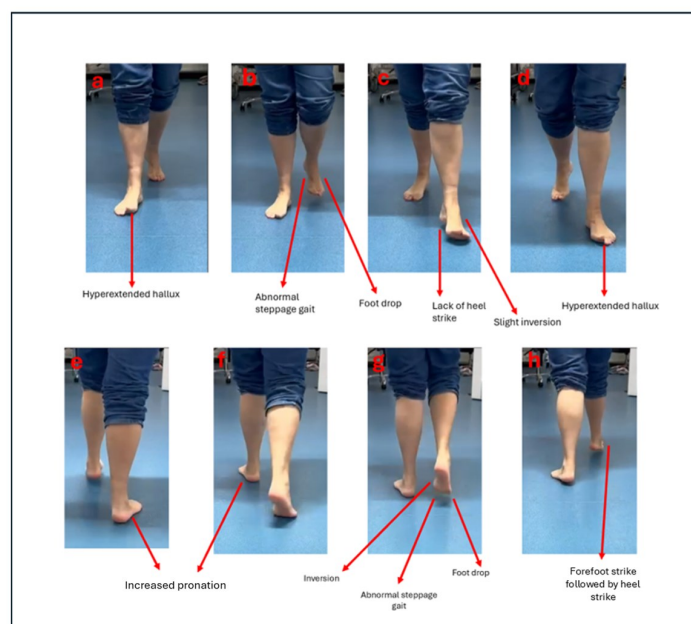


Figure 2- (a-h) Views of the patient with annotated signs indicative of Charcot-Marie-Tooth (CMT) disease during the walking cycle **a-d)** anterior view **e-h)** posterior view.

Management plan

The patient was recommended to wear the Richie Brace Dynamic Assist daily as it provides dynamic dorsiflexion assistance of up to 15 degrees which aids in patients with foot drop (10). Although the patient had an ankle equinus deformity, she lacked any severe cavovarus or planovalgus deformities and had reasonably good calf strength, therefore the Dynamic Assist was opted for over the Restricted Hinge (7).



Figure 3- Image of the Richie Brace Dynamic Assist (7).

The FFI was developed to measure the impact of foot pathology on function, initially being described for individuals with rheumatoid arthritis however its use has also been extended to orthosis outcomes due to its generalisability (11,12). The survey has demonstrated strong reliability and has proven its validity by effectively distinguishing between individuals with varying levels of foot health issues. The FFI consists of three sub-scales assessing pain which measures the severity of foot pain, disability which evaluates the degree of difficulty patients experience when performing daily tasks and activity limitation which assesses the frequency of activity limitations caused by the foot problems (11). The FFI uses the visual analogue scale to establish scores for each sub-section which is averaged to find the total FFI score, where higher scores represent worse foot health. This was chosen over the newer Revised FFI as the original FFI is more established in the literature compared to the Revised FFI (13). The patient answered the questionnaire before treatment began and was followed up after three months.

Results

Across all sub-scales of the FFI, scores improved after 3 months of using the Richie Brace Dynamic Assist. Foot pain improved from 79% to 28%, disability 52% to 17% and activity limitation improving from 40% to 11% (table 1).

Table 1- The patients overall scores for the 4 sections of the Foot Function Index (FFI).

	Pre-treatment %	Post-treatment %
Foot pain	79	28
Disability	52	17
Activity limitation	40	11

Scores are on a range of 0-100 where 0 indicates the best foot health. The questionnaire was repeated 3 months post-treatment

Discussion

This 36 year old female patient had a history of CMT disease with muscle wasting and reduced ankle reflexes. As a result, a foot drop with inversion was observed through swing requiring a high steppage gait for ground clearance which is classically seen in CMT disease (1). The inversion was the result of the reduced antagonistic action of the peronei. During late swing / early stance the hallux was hyperextended due to the lack of dorsiflexion of the ankle, in an attempt to clear the ground during this phase. It is unknown as to why only some of the muscles within the anterior and lateral compartment are affected and others such as the tibialis anterior remains unaffected however it may do with the pathophysiology of differing CMT disease types (9). The foot drop resulted in a forefoot strike, followed by heel strike and increased pronation and was felt to be contributing to her pain and callous formation.

Given that the overall foot function was affected through swing and stance, it was important to be able to improve function through both phases in order to reduce the overall foot pain. As a result, the patient was recommended the Richie Brace Dynamic Assist. This specific Richie Brace (10,14) was chosen because:

1. It has a spring loaded hinge at the ankle which actively dorsiflexes the foot and allows heel contact.
2. The medial and lateral stirrups also provide frontal plane stability reducing the inversion through swing.
3. The orthoses component, combined with the frontal plane stability of the stirrups is extremely effective at reducing pronation.
4. The hinge allows the ankle to dorsiflex through stance

The standard Richie brace (which does not have the spring loaded foot) would have been insufficient to control the foot drop. There is a fixed hinge version which reduces ankle plantarflexion or dorsiflexion. This is generally used for patients with arthritic joints as it restricts motion, which was not the aim in this case.

At three month review, the patient showed a general improvement in foot health. All sections of the FFI showed improvements and the patient expressed her general happiness with the results after using the Richie Brace Dynamic Assist. Landorf et al (15) identified that a minimal important differences for the FFI are 12 for foot pain, 7 for disability and 7 for activity limitation, where these results go far beyond these guidelines. The patient will be followed up in 6 months and again after a year to assess any changes in order to evaluate her long term foot status. AFO use has been described to be ceased due to multiple reasons such as cosmetic appearance and tolerability (16). As a result, the patient will be reviewed in six months and at one year to assess compliance and ongoing benefit

Orthoses have been described as an essential part of the rehabilitation of individuals with CMT disease complete with a rocker sole whilst being individualised to the patients needs (1). A study by Ramdharry et al (17) investigated 3 commercially available AFO's and its ability to manage footdrop in patients with CMT who had varying foot deformities. These included a, 'Footup splint,' a 'Push brace,' and the Multifit Achilles drop foot orthoses. Overall, they found that all three AFO's helped to improve dorsiflexion during the swing phase of walking but had no effect on power generation. The stiffness of certain orthoses has been identified as a possible reasoning as to why power generation improves, with stiffer orthosis providing better power results and would be interesting to investigate on future follow ups in our patient (18,19).

Although there was success for this patient in terms of pain relief and general satisfaction with her results, it is important to note that CMT disease can occur in multiple forms and present differently across patients. Originally CMT disease was classified into CMT-1 and CMT-2 however, due to the increased utilisation of next generation sequencing, more genes have been identified and linked with CMT disease, which has made it increasingly difficult to classify (20). CMT-1 is largely caused by genetic defects in either PMP22, GJB1 or MPZ which leads to Schwann cell and myelin sheath damage (21). CMT-2 represents a smaller proportion of cases however is associated with mutations in the MFN2 gene causing axonal degeneration (21). CMT-X, CMT-3 and CMT-4 are all rarer forms, presenting with different pathologies. Although pes planus is a feature described in CMT disease, it is less prevalent to see this within the adult population (22). As a result, data lacks in the appropriate management of these individuals, unlike those presenting with cavus deformities. Hoellwarth et al (22) stated that the majority of paediatric patients begin with pes planovalgus deformity however as the child ages, cavovarus deformity predominates.

Therefore, it's vital to identify specific adult patients with pes planovalgus or planus deformities in order to provide better evidence for certain AFO's that may be used in these specific cases.

Conclusion

The Richie Brace Dynamic Assist provided both pain and functional improvement in a patient with CMT disease with a pronated foot type after three months of treatment. The patient will be followed up and the FFI used in order to determine continued use and longer term results.

Conflicts of Interest

The authors declare no conflict of interest.

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