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# FUNCTIONAL KINETIC PROFILE IN CHARCOT MARIE TOOTH CARRIER

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**Abstract:** Charcot Marie Tooth (CMT) is an autosomal dominant Mendelian neurological disease. It is characterized by weakness and progressive muscle atrophy, initially in the distal muscles of the lower limbs and later in the upper limbs, sensory and motor alterations, deformities in the feet, later affecting the hands. The role of physiotherapy in patients with CMT is to maintain a safe and effective gait, minimize the manifestations of the disease and optimize the function that patients have. Objective: To verify the functional kinetic profile of individuals with CMT from the ``Associação Brasileira dos Portadores de Charcot Marie Tooth`` in different types of the disease. Method: Prospective investigative study. 17 questionnaires from volunteers of both sexes, over 18 years old, with CMT disease, were included. through an electronic questionnaire. Results: Through the data obtained, it was possible to identify the main functional kinetic changes in patients with CMT. Conclusion: Based on the results obtained through the questionnaires, the functional kinetic profile of the ABCMT member volunteers was verified. Where average age, onset of symptoms, most frequent type was related. Gait quality, balance, use of aids, tendon reflexes and degree of disability through a neurological score – adapted. onset of symptoms, most frequent type. Gait quality, balance, use of aids, tendon reflexes and degree of disability through a neurological score – adapted. onset of symptoms, most frequent type. Gait quality, balance, use of aids, tendon reflexes and degree of disability through a neurological score – adapted.
**Keywords:** Functional kinetic profile; CMT; Hereditary sensorimotor neuropathy; Physiotherapy.

## INTRODUCTION

Charcot Marie Tooth disease (CMTD) is an autosomal dominant mendelinian neurological disease. CMT is a neuromuscular disorder characterized by progressive, length-dependent degeneration of peripheral nerves, resulting in muscle weakness and atrophy in the distal limbs, feet, and hands. Onset varies from childhood to late adulthood and clinical severity varies from mild to severe among patients. Neurophysiological and neuropathological defects in motor and sensory nerves generate foot deformities, gait changes, wheelchair dependence, and sensory deficits (TIMMERMAN, 2014). About 75% of cases are caused by a duplication in the region of the PM22 gene of the peripheral myelin protein, resulting from unequal crossing over in the chromosome (KIERSZEMBAUM, 2012).

DYCK and LAMBERT (1968) defined two large groups: the first demonstrated a reduction in the median nerve conduction velocity, in addition, they also presented a nerve demyelination process on pathological examination, some with hypertrophic alterations. The second group had neuronal degeneration on pathological examination with normal conduction velocity. Therefore, CMT1 is considered a disease with demyelinating characteristics with conduction velocity < 38 m/s and CMT2 a disease with axonal characteristics with conduction velocity > 38 m/s. According to Timmerman (2014), in CMT1 myelin Schwann cells are affected, while axons suffer degeneration in CMT2. Other divisions can be based on the pattern of transmission, such as CMT3, which is also known as Dejerine-Sottas disease, where the conduction velocity is extremely slow, a combination of axonal and demyelinating neuropathy occurs, and has an autosomal dominant origin. And also CMT4 which refers to demyelinating or axonal DCMT with an autosomal recessive transmission pattern. Generally, the conduction velocity is slow < 38 m/s. (DYCK; LAMBERT, 1968).

CMTX is inherited in an X-linked dominant pattern. Inheritance is dominant if one copy of the altered gene is sufficient to cause the condition. In most cases, affected men, who have the change in their (DYCK; LAMBERT, 1968). CMTX is inherited in an X-linked dominant pattern. Inheritance is dominant if one copy of the altered gene is sufficient to cause the condition. In most cases, affected men, who have the change in their (DYCK; LAMBERT, 1968). CMTX is inherited in an X-linked dominant pattern. Inheritance is dominant if one copy of the altered gene is sufficient to cause the condition. In most cases, affected men, who have the change in their women, who have two X chromosomes. A feature of X-linked inheritance is that parents cannot pass on X-linked disease traits to their children. all daughters in men affected will have one chromosome X changed, but if they developed showed only mild symptoms of the disease. (MURFHY et al, 2012) Table 1 shows each type, the genetic profile and the proportion that occurs.

Autosomal recessive neuropathies tend to have an earlier onset (usually in early childhood) and a more severe progression than autosomal dominant neuropathies.

The clinical manifestations of CMT disease are weakness and progressive atrophy, initially in the distal muscles of the lower limbs, which may later reach the upper limbs, presenting deformity in the feet, loss of distal sensitivity, hyporeflexia or even areflexia. The first signs and DCMT symptoms appear in childhood or adolescence, with motor alterations as the main complaint. Difficulty running, jumping, and easy falls are signs of muscle weakness in distal segments. Presence of pes cavus, hammer toes, paresis of the extensor digitorum brevis and tibialis anterior muscles are the most suggestive signs for the recognition of CMT. (THOMAS et al 1997; HARDING; THOMAS, 1980; MARANHO; VOLPON, 2009).

Although rehabilitation does not eliminate neurological damage in patients with DCMT, it can act in the treatment of specific symptoms, favoring functionality. The conduct must be based on prevention and the current clinical condition of the patient. Physiotherapy, then, appears to maintain a safe and effective gait, minimize the manifestations of the disease, optimize the function that patients have, preserving the range of motion and minimizing deformities caused by muscle shortening or contracture. Thus, the objectives of physiotherapeutic treatment aim to maintain trophism and decrease muscle weakness, maintenance of range of motion, preventing or delaying deformities, in order to provide an improvement in the quality of life of the CMT patient (LONGE, 2002; OATIS 1990).

Therefore, it is the role of physical therapy to monitor patients' functional abilities, determine efficient and effective ways to carry out their daily activities, explain body mechanics in order to facilitate postural changes, teach transfer techniques to patients and caregivers. For this, it is pertinent to know the manifestations and kinesiotherapeutic characteristics of patients with DCMT, so the objective of this study is to verify the functional kinetic profile of individuals with CMT in different types of the disease in the members of the Brazilian Association of Carriers of Charcot Marie Tooth.

## MATERIAL AND METHODS

This research is of an investigative nature and was approved by the ethics committee under number 503,735 (appendix A).

It consisted of a data collection, through the questionnaire formulated by the researchers (Appendix A) and a Neuropathy Score for CMT – Adapted (Appendix B) via e-mail. Data were collected from patients who are members of the Brazilian Association of

| Name from the disease[1] | Pathology | Mode in heritage | Proportion of CMT[2] cases |
|---|---|---|---|
| CMT1 | demyelinating | autosomal dominant | 40% - 50% |
| CMT2 | axonal | autosomal dominant | 10% - 15% |
| CMT3 – intermediate form | Combination in neuropathyde myelinating axonal at the individual | autosomal Dominant | Rare |
| CMT4 | Demyelinating or axonal | autosomal recessive | Rare |
| CMTX | axonal with modifications secondary to myelin | autosomal dominate | 10 – 15% |

[1] Each of the CMT subtypes (CMT1, CMT2, CMT3, CMT4, CMTX) is mainly subdivided into genetic alterations. [From Jonghe et al 1997, Nelis et al 1999]

[2] Saporta et al [2011]

Box 1: Single gene causes of CMT hereditary neuropathy

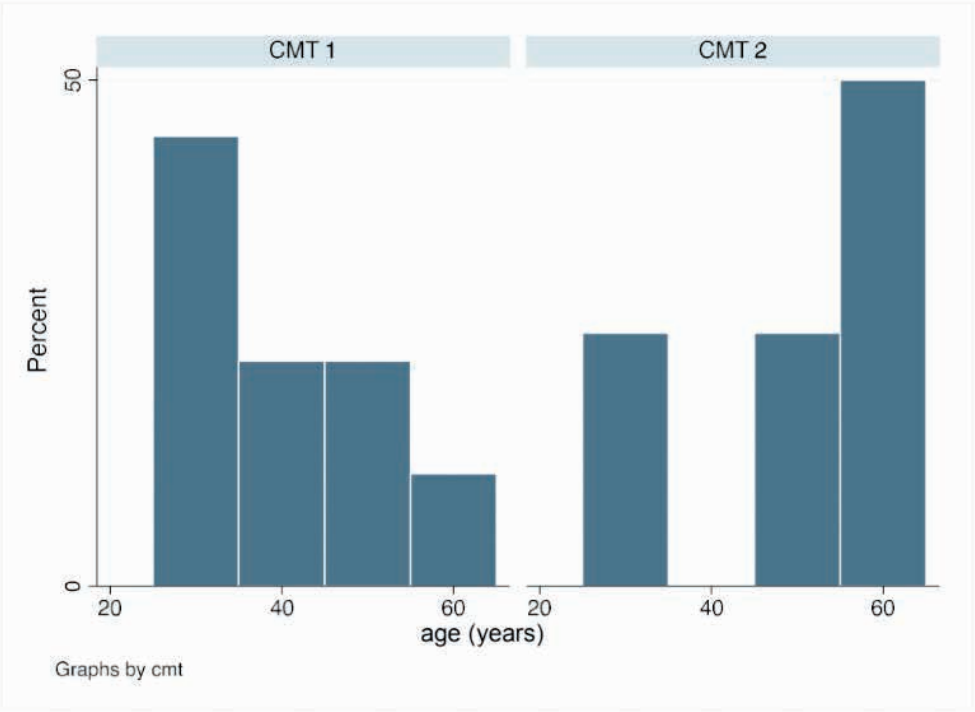


**Figure 1.** Sample age distribution by type of CMT.

Charcot Marie Tooth Carriers (ABCMT) with CMT disease, with due authorization from the President (Appendix C).

The questionnaire was developed based on evidence from the researchers' knowledge about the pathology, since there is family coexistence with it. After preparing the questionnaire, a pilot approach was carried out with a patient with DCMT who was not part of the sample, as well as the Neuropathy Score for CMT - Adapted, where it was found that both would be easy to understand and handle by patients with DCMT.

Volunteers older than 18 years of both sexes and carriers of CMT disease were included in this study. Those outside the age range and/or with some type of psychological and/or cognitive alteration were excluded. Corresponding data were collected as follows: The questionnaire and the CMT Neuropathy Score - adapted were sent to the president of ABCMT based in Ribeirão Preto, SP, who forwarded it to 40 members with CMT disease. There was no direct contact with the sample. For socioeconomic and cultural reasons, only 17 questionnaires were answered and 15 Neuropathy Score for CMT - Adapted, thus reducing the proposed number of volunteers for this study.

The questionnaire was formulated by the authors themselves with vocabulary that was easy for the patients to understand. Data were collected, such as the age at which the first symptoms appeared, as these symptoms appear in the first decade of life. Type of CMT, presence of difficulty walking, use of some aid, use of orthosis, use of prosthesis, tendon reflexes, balance, presence of deformities, performance of physiotherapeutic treatment, thus being able to trace the functional kinetic profile according to the response found in each questionnaire.

The volunteers were also asked about sensitive symptoms, painful and vibratory sensitivity, motor symptoms in both limbs, thus being able to observe where the greatest difficulties are found, contributing to form the disease profile of CMT. These data were collected using an adapted CMT Neuropathy Score.

Data analysis was performed descriptively and statistical tests were used to describe the results.

## RESULTS

17 individuals with CMT answered the questionnaire, with a mean age of 41, standard deviation (SD) of 12.7 years and median of 38. With regard to gender, 70.6% were female and 29.4% male. Among the evaluated individuals, 64.3% had a diagnosis of CMT type 1 (demyelinating), 28.6% CMT type 2 (axonal) and only 7.1% (1 individual) CMTx (axonal with secondary modifications to myelin).

In Figure 1, we have the age distribution of individuals with CMT 1, with a mean of 38.8, SD of 11.9 and a median of 37. In contrast, individuals with CMT 2 had a mean age of 49.5, with a SD of 16.6 and median= 53.5.

The duration of symptoms was on average 12 years (SD= 12.8; median= 8.5). Figure 2 demonstrates the distribution of symptom duration between types of CMT1 and 2, we observe that CMT 1 had less variability, with a mean duration of symptoms of 9.5 years (SD=7.4; median=8.5), however, for type 2 the mean time was 23.8 years and SD 19.8 (median= 23).

Of the total, only 1 individual did not walk. Among the main functional kinetic changes in the sample, 58.8% reported limping, weakness was observed in 52.9% and pain in 47%. Twenty-three percent said they needed help to walk, and 11.8% said they used prostheses and orthoses. Of the total, 93.3% reported having lost balance at some point.

With regard to tendon reflexes, in the

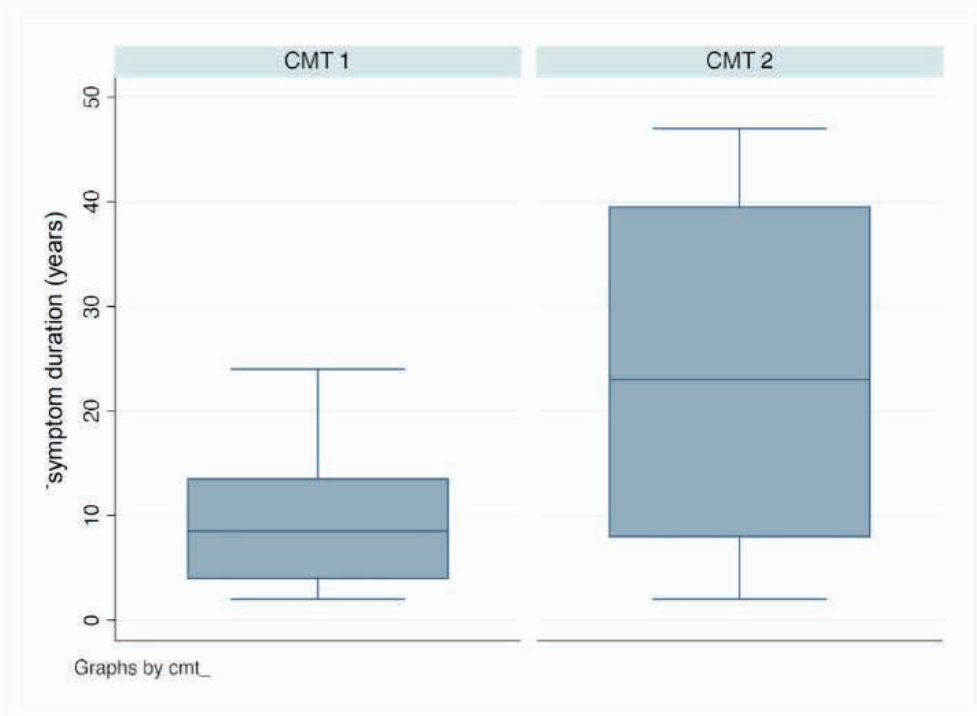


**Figure 2**. Boxplot graph of the duration of symptoms between individuals with CMT type 1 and 2.

| | CMT 1 (n= 8) | | | CMT 2 (n= 3) | | | CMTx (n= 1) | | |
|---|---|---|---|---|---|---|---|---|---|
| | Average | Median | **Min – Max** | Average | Median | **Min – Max** | Average | Median | **Min – Max** |
| **Neurological Score (scale from 0 to 100)** | 26 | 25 | 15 - 50 | 38 | 25 | 25 - 65 | 30 | 30 | - |

**Table 1.** Neurological score results (scale from 0 to 100) for CMT type 1, type 2 and CMTx in 12 subjects.

| **Symptoms** | **CMT 1 (n= 8)** | | **CMT 2 (n= 3)** | | **CMTx (n= 1)** | |
|---|---|---|---|---|---|---|
| | **n** | **%** | **n** | **%** | **n** | **%** |
| **sensitives** | | | | | | |
| Absent | 01 | 12.5 | 0 | 0 | 0 | 0 |
| Symptoms limited to the toes | 01 | 12.5 | 0 | 0 | 0 | 0 |
| They extend up to and may include the ankle | 03 | 37.5 | 02 | 67 | 0 | 0 |
| They extend up to and may include the knee | 03 | 37.5 | 01 | 33 | 01 | 100 |
| Stretch above the knee | 0 | 0 | 0 | 0 | 0 | 0 |
| **Upper limb engines** | | | | | | |
| Absent | 02 | 25 | 0 | 0 | 0 | 0 |
| Difficulty with buttons/zippers | 05 | 62.5 | 02 | 67 | 01 | 100 |
| Inability with buttons/ zippers but you can write | 01 | 12.5 | 0 | 0 | 0 | 0 |
| No it achieves to write or to use keyboard | 0 | 0 | 01 | 33 | 0 | 0 |
| Closely in the arms | 0 | 0 | 0 | 0 | 0 | 0 |
| **Upper limb engines** | | | | | | |
| Absent | 03 | 37.5 | 01 | 33 | 0 | 0 |

| | | | | | | |
|---|---|---|---|---|---|---|
| Stumbles, catches toes, throw your feet | 04 | 50 | 01 | 33 | 0 | 0 |
| AFO at the Minimum in 1 leg or ankle support | 01 | 12.5 | 0 | 0 | 0 | 0 |
| Cane, walker, ankle surgery | 0 | 0 | 01 | 33 | 0 | 0 |
| Wheelchair most of the time time | 0 | 0 | 0 | 0 | 0 | 0 |

| **sensitivity** | **CMT 1 (n= 9)** | | **CMT 2 (n= 4)** | | **CMTx (n= 1)** | |
|---|---|---|---|---|---|---|
| | **n** | **%** | **n** | **%** | **n** | **%** |
| **painful** | | | | | | |
| Normal | 04 | 50 | 01 | 33 | 0 | 0 |
| Reduced in toes/hands | 03 | 37.5 | 01 | 33 | 01 | 100 |
| Reduced until It is he can include O ankle/wrist | 01 | 12.5 | 01 | 33 | 0 | 0 |
| Reduced to and may include the knee/ elbow | 0 | 0 | 0 | 0 | 0 | 0 |
| Reduced above the knee/elbow | 0 | 0 | 0 | 0 | 0 | 0 |
| **vibrating** | | | | | | |
| Normal | 02 | 25 | 0 | 0 | 0 | 0 |
| Reduced in toes/hands | 04 | 50 | 02 | 67 | 01 | 100 |
| Reduced ankle/wrist | 02 | 25 | 01 | 33 | 0 | 0 |
| Reduced at the knee/elbow | 0 | 0 | 0 | 0 | 0 | 0 |
| Reduced above the knee/elbow | 0 | 0 | 0 | 0 | 0 | 0 |

**Table 2.** Neurological score of individuals with CMT type 1, type 2 and CMTx.

patellar region, 29.4% responded that they had abolished reflexes, 41.2% had decreased them, and 29.4% did not know how to respond. With regard to the Achilles reflexes, 35.3% of the individuals reported that they were reduced, 29.4% abolished, and 35.3% of the sample did not know how to answer the question.

Of the evaluated individuals, most of the deformities observed were 75% in the feet, 3 (25%) being unilateral and 9 bilateral (75%) and 50% in the hands, with half unilateral and half bilateral.

Regarding the Neurological Score (0 to 100), we found a mean of 26.3 (SD= 12.2 and median= 32.5) for individuals with CMT 1 and, for individuals with CMT 2, mean of 38.3 (SD=23 and median=25). These results are also shown in Table 1.

Regarding the scale items, the sensory symptoms in most individuals extend to the ankle (40%), 26.7% are limited to the toes, 26.7% include the knee and 6.7% reported no sensitivity.

Motor symptoms of the upper limbs, most reported having difficulty with buttons and zippers (66.7%), 13.3% cannot write or use a keyboard, 13.3% had absent symptoms and 6.7% were unable to use zippers, but they can write. In table 2 we have the distribution of these data in relation to each type of DCMT.

With regard to the lower limbs, 57.1% stumble, pinch their fingers and throw their feet, 28.6% have no symptoms, 7.1% have an orthosis or prosthesis on at least one leg and 7.1% use a cane, walker and ankle surgery.

Most individuals reported normal pain sensitivity with 46.7%, 40% reported it being reduced in the fingers or toes and 13.1% reduced to the hands or feet including the wrist or ankle. With regard to vibration sensitivity, 53.3% were reduced in the fingers, 20% to the wrists or ankles, 20% normal and 6.7% reduced above the elbow or knee.

When physiotherapy treatment was verified, 76.5% of the sample reported having performed it, but stopped with the treatment, 17.7% did not undergo any type of physiotherapy, and only 5.9% undergo treatment with assistance.

## DISCUSSION

In the present study, the average age of the sample was 41 years, with regard to gender, there was a predominance of females over males.

Regarding the diagnosis, a higher prevalence of CMT type 1 was observed, in relation to the others that were presented in this study. Barisic et al. (2008) aimed to discuss the clinical and neurophysiological characteristics of different types of CMT. In that study, it was reported that the type 1 CMT form is the most frequent form, corresponding to approximately 60% of cases. Type 2 DCMT is the second most frequent, with approximately 15% of the proportion of cases according to Saporta et al, (2011). Data similar to those found in this study.

Regarding the age at which the first symptoms started, there was a significant difference between CMT type 1 and CMT type 2. Muglia et al, (2001) reported that, in the case of CMT2, the age of onset is quite variable and often difficult to determine. established, and the onset of symptoms can be observed up to the fifth decade of life. Pareyson and Marquesi (2009) described the onset of symptoms in the first decade of life, which is very common in CMT1. In this study, it was observed that the average age at which the first symptoms begin between the two most frequent types, there was then a similarity to the previously mentioned studies.

Of the evaluated individuals, a high rate of difficulty in walking was observed, with claudication being the most cited, followed by weakness and pain. In addition, lack of balance appeared in most of the volunteers.

According to Meningroni et. al. (2009) lameness and weakness may be associated with weakness of the Tibialis Anterior muscle, high prevalence of pes cavus and difficulty in performing dorsiflexion, which is an important characteristic for normal gait and safe. Padua et al. (2008) reported that pain is due to musculoskeletal deformities often found in CMT. Regarding the imbalance, Maranho and Volpon (2009) claimed that it was associated with the weakness of the Peroneus Brevis muscle, which does not balance the inverting strength of the Tibial Posterior muscle, and the weakness of the Tibial Anterior muscle, with relative preservation of the strength of the Peroneus Longus muscle and Sural Triceps. There is relative preservation of the strength of the Extensor Hallucis Longus muscle, which starts to act as the ankle dorsiflexor when the Tibialis Anterior muscle is weakened, causing great instability.

As for the use of aids, orthoses and prostheses, few patients were found to use them. According to Pereira et. al. (2012), its importance in patients with DCMT becomes relevant because it promotes improvements in balance reactions and gait performance, due to the large number of musculoskeletal deformities, which cause numerous functional changes. Thus, carrying out treatment through the use of assistive equipment can minimize inadequate movement synergies and optimize function in these patients. For Holmes and Hansen (1993), appropriate orthoses help to distribute the weight on the soles of the feet and compensate for the position of a hindfoot varus, as well as appropriate shoes accommodate hammer toes.

The responses obtained on tendon reflexes were analyzed, it was found that the majority when it came to the patellar reflex, presented hyporeflexia, as well as the Achilles tendon reflex. Areflexia was also considered by a large part of the sample. Barisic et al, (2008) claimed that the reduction and even the abolition of reflexes are the rule, although in some forms of CMT one can find exalted reflexes and even the presence of Babinski's pyramidal sign, but spasticity is not found. The answers found in this question were subjective, as there was no contact with a professional in the area to carry out the evaluation.

Maranho and Volpon (2009) described that in DCMT the presence of deformities is quite common, and the origin of most deformities is related to the imbalance of the intrinsic and extrinsic muscles of the foot, with very diverse patterns. Harding and Thomas, (1980) explained a possible relationship with the paresis of some muscles, such as the Extensor Brevis of the fingers, Tibialis Anterior, Dorsal Interossei of the hand and the Thenar region. In the present study, a higher rate of foot deformities was found, bilaterally. Being the most cited presence of pes cavus. As for the hands, there was a lower prevalence of deformities, the most cited being muscle atrophy.

Regarding the items of the neurological score, it was observed in this study that the DCMT presents less elevated sensory symptoms than the motor alterations. As well as the mild degree of disability with activities of daily living. Gemignani et. al. 2004 reported that it is not common for CMT patients to present positive sensory symptoms. Kleyn, Dyck, (2005) stated that sensitive alterations may appear later than motor manifestations, making the diagnostic definition of DCMT difficult. In the opinion of Barisic et. al. 2008 most of the time, although it is difficult to precisely define which distal muscles are affected in the upper limbs, difficulties in daily activities in relation to manual tasks can be mentioned. Data similar to those obtained in this study.

Neves and Kok, (2011) concluded in their study that all forms of CMT cause length-

dependent axonal degeneration, which is a factor responsible for motor and sensory deficits. He also related that the loss of a clinically evaluated function is better related to axonal degeneration than to myelin damage. He also emphasized that, among the various ways already described to assess the degree of disability in CMT, the CMT neuropathy score is the one that seems closest to translating the real damage caused by CMT.

Another issue relevant to the present study is the physiotherapeutic treatment, where the vast majority of volunteers had already undergone physiotherapeutic treatment, however, interrupted it, reporting the progression and worsening of the atrophy. This may be related to the type of conduct taken by the professional physiotherapist. Kilmer et al (1994) report that the benefits and risks of muscle strengthening exercises for people with neuromuscular diseases, including patients with CMT, have been discussed in the literature, since there are studies reporting improvement in muscle strength with exercises of resistance, as well as reports of increased muscle weakness from overuse of weak muscles.

Another justification presented was the lack of knowledge of the professional physiotherapist about DCMT, causing insecurity in patients with the disease. Thus, the importance of further research is observed, emphasizing the best conduct, aiming at benefits to patients and better basis and support to the professional.

## CONCLUSION

Based on the results obtained through the 17 questionnaires, the functional kinetic profile of the ABCMT member volunteers was verified. Being the average age of 41 years, with a predominance of females. There was a higher prevalence of type 1 CMT (64.3%), followed by type 2 CMT (28.6%). The average age of individuals with CMT 1 was 38.8 years. On the other hand, CMT 2 patients had an average of 49.9 years.

The symptoms started around 12 years of age in general among the types.

The symptomatology was 9.5 years in CMT1 and 23.8 years in CMT2.

With regard to functional symptoms, it was found that the main changes are claudication, weakness and pain when walking, as well as loss of balance (93.3%) in both ambulation and orthostasis.

According to the tendon reflexes, hyporeflexia was confirmed both in the patellar region and in the aquile region. A higher rate of deformity was observed in the lower limbs bilaterally. Not excluding upper limbs with present deformity both unilaterally and bilaterally (50% each). Regarding sensory symptoms, the sample has symptoms that extend to and may include the ankle. As well as having difficulty with buttons/zippers in MMSS. In MMII, the profile is tripping, pinching your fingers and throwing your feet. Pain sensitivity is preserved in the specimen and vibratory sensitivity is reduced in the fingers/toes.

Based on this profile, we verified the importance of tracing a functional kinetic profile in a theoretical and practical way to gain knowledge and relate the activities of daily living of these individuals, thus being able to elaborate the most appropriate intervention possible.

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## ATTACHMENT

### ANNEX A

CENTRO DE ENSINO SUPERIOR DOS CAMPOS GERAIS - CESCAGE/PR

Plataforma Brasil

**PARECER CONSUBSTANCIADO DO CEP**

**CONSUBSTANTIATED OPINION OF THE CEP**

**RESEARCH PROJECT DATA**

**Title of the Research:** CIKETICOFUNC" PROFILE IN CHARCOT MARIE TOOTH

**Researcher:** PATRICIA CAMARGO

Thematic Area:

Version: 1

CAAE: 25429113 9 0300.5215

Proponent Institution: CESCAGE CENTRO DE ENSINO SUPERIOR DOS CAMPOS GERAIS - ME

Main Sponsor: Own Funding

OPINION DATA

Opinion Number 503.735

Date of Report: 11/122013

Project Presentation:

This research aims to verify the functional kinetic profile of individuals with CMT in the different types of the disease in the members of the Brazilian Association of Charcot Marie Tooth Carriers

Objective of the Research

To verify the kinetic functional profile of individuals with CMT in the different types of the disease in the members of the Brazilian Association of the Carers of Charcot Marie Tooth

Evaluation of the Risks to Benefits:

There are no risks, because the individual carrying this pathology will only be observed.

The benefits are of great value, since the individual with this pathology will be accompanied, where the researcher will evaluate how the progression of this disease occurs, and will be able to find better alternatives for the treatment of this disease.

Comments and Considerations about the Research:

There are no

Consideration about the Terms of Mandatory Submission:

Meets the prerequisites

Bairro: Uvaranas CEP: 84.030-000

UF: PR Município: PONTA GROSSA

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CENTRO DE ENSINO SUPERIOR DOS CAMPOS GERAIS - CESCAGE/PR

Opinion confirmation: 503,735

**Recommendations:**
Send partial and final report to the committee.

**Conclusions or Pending and List of Inadequacies:**
None

**Status of Opinion**
Approved

**Needs Appreciation of CONEP:**
No

**Final considerations at the discretion of CEP:**
After deliberation of the collegiate maintained approval of the project by the rapporteur

PONTA GROSSA, 20 de Dezembro de 2013

Signed by:
**Sylvio Reynaldo Schieder**
(Coordinator)

**Endereço:** Av. Gen. Carlos Cavalcanti, 8000
**Bairro:** Uvaranas **CEP:** 84.030-000
**UF:** PR **Município:** PONTA GROSSA
**Telefone:** (42)3219-8039 **Fax:** (42)3219-8001 E-mail: cep@cescage.edu.br

## ANNEX B

| Neuropathy Score for CMT – ADAPTED | | | | | |
|---|---|---|---|---|---|
| parameters | score | | | | |
| | 0 | 1 | 2 | 3 | 4 |
| Symptoms sensitives | Absent | Symptoms limited to the fingers of the feet | extend to and can include the ankle | stretch out until and can include the knee | stretch out above of knee |
| Symptoms engines legs | Absent | trips, arrest the fingers of feet, throw feet | AFO at least on 1 leg or support ankle | Walking stick, walker, surgery ankle | Chair of bigger wheels part of time |
| Arms | Absent | Difficulty with buttons / zippers | Inability with buttons/zippers but can to write | Can not write or use keyboard | Proximally in the arms |
| sensitivity painful | Normal | Reduced on the fingers of hands/feet | reduced to and can include wrist/ankle | reduced to and can include elbow/knee | Reduced above of elbow/ knee |
| sensitivity vibratory | Normal | Reduced on the fingers of hands/feet | Reduced in wrist/ ankle | Reduced in elbow/ knee | Reduced above of elbow/ knee |

Caption: AFO = foot ankle orthosis; Source: Shy et al., 2005

**ANNEX C**

ABCMT Associação Brasileira dos Portadores de Charcot Marie Tooth
Rua Gedeon Alves Feitosa 167 – Jd. Independência.
Cep: 14.076.240 / Ribeirão Preto – SP
Contato: 16 3626 9248/ 16 8206 2684/ 16 9323 6365
CNPJ 13. 815 481 / 0001 – 35

Ângela Mérici Alves

## DECLARATION´

I. ÁNGELA MERICI ALVES. President of the Brazilian Association of Pruners of Charco' Marie Tooth - ABCMT authorize the development of the research project 'FUNCTIONAL KINETIC PROFILE OF THE MARIE TOOTH CHARCOT", by the student of the Physiotherapy course of the Centro de Ensino Superior dos Campos Grais - CESCAGE. Ponta Grossa - PR, Andressa Camargo e Silva, to be developed with members of the ABCMT. located at Rua: Gedeon Alves Feitosa 167 -Jardim Independência. Zip Code: 14.076 - 240 / Ribeirão Preto - SP

Ribeirão Preto, November 18, 2023

Ângela Merici Alves
ABCMT President
Id card:10596639-3

**Term of authorization for use of the questionnaire**

**I,** Angela Merici Alves, CPF (social security number) 02973445876_. Id card: 105936393, after knowing and understanding the objectives, methodological procedures, risks and benefits of the research, as well as being aware of the need to fill out this questionnaire and/or testimonial specified in the Informed Consent Form, I hereby AUTHORIZE The researcher (Andressa Camargo e Silva) of the research project entitled

"KINETIC FUNCTIONAL PROFILE OF THE CHARCOT MARIE

TOOTH" to carry out the necessary questionnaire and to collect my testimony without any financial burden to any of the parties.

At the same time, I authorize the use of this information for scientific and study purposes (books, articles, slides, and transparencies) on behalf of the researcher of the above research, in accordance with the provisions of the laws that safeguard the rights of children and adolescents (Child and Adolescent Statute - ECA Law Number: 8.0891 1990), the elderly (Elderly Statute Law No. 10.741/2003) and people with disabilities (Decree Number: 3.298/1999, as amended by Decree Number: 5.296/2004).

Ribeirão Preto, November 18, 2023

Assinatura do participante/representante legal

Signature of participant/legal representative

Andressa Camargo e Silva - pesquisadora principal

Andressa Camargo e Silva – lead researcher

## APPENDIX

### APPENDIX A

**CMT Questionnaire**

**Name (initials):**
**Age: _____ Weight: _____ Height: _____ Sex: _____**
**How old were the first symptoms?**_____
**At what age was the disease diagnosed? What is the type of CMT?**_____
CMT1 ( ) CMT2 ( ) CMT3 ( ) CMT4 ( ) CMTX ( )

**Are there other cases in the family of CHARCOT MARIE TOOTH Disease? If so:**
How many?_____
What type?_____
Degree of kinship:_____

**Do you use any medication? If yes, which ones?**

_____

_____

**Do you wander?**(walks)
Yes ( ) No ( )

**Do you have difficulty ambulating (walking)?**
( ) Claudica (limp) ( ) feel pain ( ) feels weakness ( ) other:_____

**Do you use any help? If yes, which one?**
( ) crutch ( ) wheelchair ( ) walker ( ) other:_____

**Do you use an orthosis (splint)? If yes, where?**
( ) Knee ( ) ankle ( ) foot ( ) Other:_____

**Do you have any type of prosthesis? Which?**
( ) pin ( ) plates ( ) screw ( ) other:_____Where?_____

**Did you have any kind of surgery? Which?**
( )Stretching ( )Fixation ( ) Botox ( )Other:_____

**Does it have tendon reflexes (immediate and involuntary movement of a limb after a stimulus)? If yes, how is it?**
Patellar (knee)
( ) Decreased (when stimulated, he perceives movement, but it is slight)
( ) Exalted / increased (when stimulated, it perceives movement and it is exaggerated)

( ) Abolished/absent (when stimulated there is no movement) Achilles (ankle)
( ) Decreased (when stimulated, he perceives movement, but it is slight)
( ) Exalted / increased (when stimulated, it perceives movement and it is exaggerated)
( ) Abolished/absent (when stimulated there is no movement)

**Do you have any balance changes? If yes, when?**

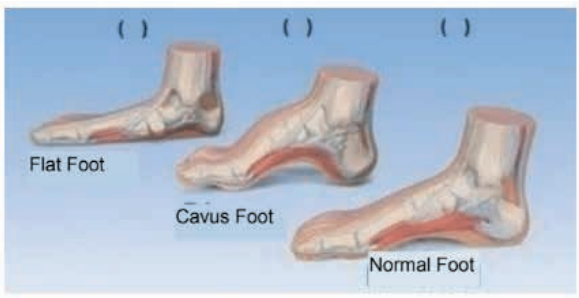
_____

**When you walk, do you notice a change in the distance between your legs?**
( ) increased ( ) decreased ( ) normal

**Do you have any reduced musculature? If yes, where?**

_____

Do you have any changes in your feet? If yes, which one?




**Do you have tremors? If yes, where?**
( ) feet ( ) legs ( ) thigh ( ) another place:_____

**Do you have any kind of pain? Where?**
Yes ( ) no ( )

**Do you have any kind of deformity? Where?**
Yes ( ) no ( )
D hand ( ) Foot D ( )
Hand E ( ) Foot E ( )

**Do you practice physical activity? If yes, which one?**

_____
How many times a week?_____

**Have you ever had or are you undergoing physiotherapeutic treatment?**
( ) No
( ) Yes, but I stopped. How much time?_____
What is the reason?_( ) Yes, I continue How long?_____
How many times a week?_____

What is your opinion about the physiotherapeutic treatment in relation to the progression of the disease?

_____

_____

_____