Available online at
ScienceDirect
www.sciencedirect.com
Elsevier Masson France
EM|consulte
www.em-consulte.com
Annals of Physical and Rehabilitation Medicine 57 (2014) 587–599
ANNALS OF PHYSICAL AND REHABILITATION MEDICINE

Original article / Article original

The motor function measure to study limitation of activity in children and adults with Charcot-Marie-Tooth disease

Utilisation de la mesure de fonction motrice pour étudier des limitations d'activités de patients adultes et enfants atteints de la maladie de Charcot-Marie-Tooth

L. Allard [a,*], G. Rode [a], S. Jacquin-Courtois [a], M.C. Pouget [a], P. Rippert [b,c], D. Hamroun [d], I. Poirot [b], C. Bérard [b], C. Vuillerot [b,e,f,g] le groupe d'étude CMT MFM[1]

[a] *Service de médecine physique et réadaptation, hôpital Henry-Gabrielle, hospices civils de Lyon, 20, route de Vourles, 69230 Saint-Genis-Laval, France*
[b] *L'Escale, service central de rééducation pédiatrique, hôpital femme Mère–Enfant, hospices civils de Lyon, 59, boulevard Pinel, 69677 Bron cedex, France*
[c] *Pôle information médicale évaluation recherche, hospices civils de Lyon, 69003 Lyon, France*
[d] *Direction de la recherche et de l'innovation, centre hospitalo-universitaire de Montpellier, 34000 Montpellier, France*
[e] *Université de Lyon, 69000 Lyon, France*
[f] *Université Lyon I, 69100 Villeurbanne, France*
[g] *CNRS, UMR 5558, laboratoire de biométrie et biologie évolutive, équipe biostatistique santé, 69310 Pierre-Bénite, France*

Received 16 December 2013; accepted 8 September 2014

Abstract

Objective. – To study the applicability and responsiveness of the motor function measure (total score and sub-scores D1, D2 and D3) in patients with Charcot-Marie-Tooth disease.
Patients and methods. – Two hundred and thirty-three patients aged 4–86 years were included in the descriptive study. Scores and sub-scores were analyzed by age and by disease subtypes. Sensitivity to change (responsiveness) was estimated in patients having had at least two evaluations with at least six months between the first and the second.
Results. – Motor function measure scores decrease with age, especially sub-scores D1 and D3. There were no significant differences between the scores according to type of Charcot-Marie-Tooth disease. The scores were significantly higher for ambulatory than for non-ambulatory patients. Significant responsiveness was demonstrated only in type 2 Charcot-Marie-Tooth disease.
Discussion/conclusions. – Our results suggest that, especially for D1 and D3 sub-scores, the motor function measure is a reliable and valid outcome measure that can be usefully applied in longitudinal follow-up. Studies of longer duration could demonstrate its responsiveness in other Charcot-Marie-Tooth disease subtypes.

Keywords: Charcot-Marie-Tooth disease; Motor Function Measure; Activity limitation; Neuromuscular disease; Functional scale

Résumé

Objectif. – Étudier l'applicabilité et la sensibilité au changement du score total et des trois sous-scores, D1, D2 et D3 de la mesure de fonction motrice chez des patients atteints de maladie de Charcot-Marie-Tooth.
Patients et méthodes. – Étude descriptive sur 233 patients âgés de 4 à 86 ans. Les scores ont été analysés en fonction de l'âge et du type de la maladie. Les sensibilités au changement ont été étudiées chez les patients ayant eu au moins deux évaluations espacées d'au moins six mois.
Résultats. – Les scores de la Mesure de Fonction Motrice diminuent avec l'âge, surtout les sous-scores D1 et D3. Il n'existe pas de différence significative en fonction du type de la maladie. Les scores des patients non ambulants sont significativement plus faibles que ceux

* Corresponding author.
E-mail address: laurenceallard@free.fr (L. Allard).
[1] Groupe d'étude constitue de médecins, kinésithérapeutes et ergothérapeutes français et suisse ayant fourni des données MFM de patients CMT.

http://dx.doi.org/10.1016/j.rehab.2014.09.005
1877-0657/

des patients ambulants. La sensibilité au changement est significative chez les Charcot-Marie-Tooth type 2.
Conclusion. – La mesure de fonction motrice est une échelle applicable, surtout D1 et D3, au suivi des patients atteints de la maladie de Charcot-Marie-Tooth de façon longitudinale. Sa sensibilité au changement nécessite d'être confirmée en utilisant des durées d'étude plus longues compte tenu de l'évolutivité de la maladie.

Mots clés : Maladie de Charcot-Marie-Tooth ; Mesure de Fonction Motrice ; Limitation d'activités ; Maladie neuromusculaire ; Échelle fonctionnelle

1. English version

1.1. Introduction

Charcot-Marie-Tooth disease (CMT) is one of the most frequent hereditary neuromuscular diseases (NMD), with a prevalence in France of 1 out of 2500 persons [1]. In fact, CMT is a group of diseases characterized by damage to the nerves of the arms and the legs (peripheral nerves) and presenting considerable genetic heterogeneity, with more than 30 potentially responsible genes [2–5]. Patients suffering from CMT are categorized in sub-groups according to nerve damage and mode of transmission (CMT1: demyelinating form, autosomal dominant; CMT2: axonal form, autosomal dominant or recessive; CMT4: demyelinating form; autosomal recessive; CMTX: form related to chromosome X and IAD: intermediate autosomal dominant) [4] and present highly variable clinical pictures (peroneal muscle atrophy, damaged intrinsic hand muscles, areflexia, deformation of the feet...). Disease development is often slowly progressive and age at symptom onset greatly varies from one individual to the next [7–9].

Even though no medical CMT treatment is currently available, studies carried out on animal models have yielded promising results [2,10–12] and trials have been or will soon be conducted in humans. Moreover, the effects in humans of ascorbic acid, progesterone and its derivatives, neurotrophine 3 and curcumin [2,3,13,14] have been studied. That much said, precise study of the effects in humans of therapeutic products necessitates valid, sensitive and reproducible assessment tools. From a clinical standpoint, functional assessment both improves long-term patient follow-up and enhances knowledge of the natural development of the disease. More specifically, functional assessment can help to anticipate and forestall some consequences of the disease through appropriate management and adequate technical support.

In 2010, the 168th ENMC international workshop provided an occasion for a review of the available CMT assessment tools [6]. Among these tools, the most widely used are the CMT neuropathy score (CMTNS) [15], which is the only CMT-specific measurement, and the neuropathy impairment score (NIS) [16]. Both of them have been validated on patients of at least 10 years of age. While the CMTNS takes into account the findings of the neurological exam, the results of electromyogram testing, sensitivity and muscle strength [17], the NIS scale is a standard scale that evaluates 24 muscle groups, five osteo-tendinous reflexes and disturbed sensation, which is assessed on the dorsal side of the last phalanx of the index finger and the great toe. Only in cases of CMT1A and CMTX has the course of CMT development been studied using the CMTNS and the NIS. The two scales have been shown to effectively detect CMTA change and progression, and the authors recommend study of the development of the other types of CMT using this tool [15].

As CMT onset generally occurs during the first decade of life, a scale known as the CMT Pediatric Scale (CMTPedS) has been developed [18]; it is well-tolerated, reliable and reproducible. However, since it is usable only for children, it does not allow for longitudinal follow-up after transition in the adult.

The motor function measure (MFM) is a precise scale that has been validated for most cases of NMD. It objectifies the repercussions of muscle weakness on motor functioning and is appropriate with regard to most neuromuscular diseases, whatever their level of severity. The scale exists in two versions, one of which consists in 32 items and has been validated for NMD patients aged from six to 60 years (MFM-32), while the other comprises 20 items (MFM-20) and has been validated for children of two to six years of age [19–21]. Validation studies of MFM-32 and MFM-20 have led to the categorization of three motor functions:

- D1: standing position and transfers (13 items for MFM-32 et 8 for MFM-20);
- D2: axial and proximal motor function (12 items for MFM-32 et 8 for MFM-20);
- D3: distal motor function (seven items for MFM-32 et 4 for MFM-20).

The proof-of-concept MFM-32 study, which was conducted in a population of 303 NMD patients of whom 35 were suffering from peripheral neuropathy, successfully demonstrated the robust metrological properties of the tool [20], and later studies conclusively demonstrated its validity and responsiveness in the different groups of NMD patients [19,22–24].

One advantage of the MFM-32 is that contrarily to the CMTNS, NIS and CMTPedS scales, it is applicable to patients aged from six to 60 years; moreover, it does not take into account the data provided by an electromyogram (EMG), which remains an invasive test that is difficult to carry out systematically, during each consultation.

The objective of this work is to study the applicability and, more particularly, the responsiveness or sensitivity to change (for the total score and the three sub-scores, D1, D2 and D3) of the MFM scale in patients with CMT.