Rapid communication

Charcot-Marie-Tooth disease and rehabilitation: a perfect match

Reabilitação e doença de Charcot-Marie-Tooth: um casamento perfeito

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Charcot-Marie-Tooth disease (CMT), a hereditary motor and sensory neuropathy, is one of the most common inherited neurological disorders, affecting approximately 1 in 2,500 people in the world. A typical feature includes weakness of the foot and lower leg muscles, which may result in foot drop and a high-stepped gait with frequent tripping or falls.[1]

Onset of symptoms is more often in adolescence or early adulthood, but some individuals develop symptoms in mid-adulthood. The severity of symptoms varies greatly among individuals and even among family members with the disease. Progression of symptoms is gradual. Pain can range from mild to severe, and some people may need to rely on foot or leg braces or other orthopedic devices to maintain mobility. Although in rare cases, individuals may have respiratory muscle weakness.[2]

New approaches and techniques for rehabilitation of patients with CMT emerged lately, some consolidated, others lacking evidence. The role of physiotherapy aims to maximize functional ability and minimize secondary complications through movement rehabilitation within a context of education and support for the whole person. It is worth mentioning that as any neurological disease of progressive nature, there is a time when the physiotherapist is only able to promote adaptations, guidelines and logically manage losses in gait patterns, and alleviating the risk of falls.

Recently, researchers have created a wearable tactile feedback system, developed previously for sensory augmentation of prosthetic limbs, and adapted for individuals with Charcot-Marie-Tooth disease and other peripheral neuropathies. Patients were assessed both for their abilities to perceive tactile stimuli, and for the effect of tactile biofeedback on their gait. Preliminary data indicate that participants could localize tactile stimuli and make modifications on their gait. However, the effect of feedback on gait was highly variable from subject to subject, requiring further investigation.[3]

Currently, our group examined the immediate effects of the use of ankle-foot orthosis (AFO) in kinematic gait and balance in patients with CMT. Nine subjects were evaluated by the Tinetti scales and Dynamic Gait Index (DGI) and the kinematic parameters of gait through kinematics. The evaluations were performed before and during use of orthoses. Significant change was observed in DGI scale during

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the use AFO ($p < 0.05$). The use of AFO promoted immediate effects on gait kinematics and in the balance reactions. The results suggest that the use of OTP is an effective strategy to stabilize the ankle as well as to prevent foot drop in patients with CMT. We alert that the use of partial body weight support for those patients who walk with extreme difficulty can and should be a strategy to be attempted.

The hydrotherapy associated or not with conventional physiotherapy on the floor is also a treatment strategy, because it minimizes the risk of falls, reduces the gravitational force and favors the execution of gait patterns. Authors investigated the effects of hydrotherapy associated with kinesiotherapy in patients with CMT through Timed Up and Go, gait speed, the Six-Minute Walk Test and Berg Scale. A total of 21 sessions is conducted with two weekly visits of 1 hour (pre-established protocol). After the practice of therapeutic exercises the patient was able to go up and down stairs more easily, the postural instability was attenuated and the distance and gait speed also experienced significant gains.[5]

A problem frequently found in this group of patients are joint contractures. They may contribute to increased disability due to decreased motor performance, mobility limitations, reduced functional range of motion, loss of function for activities of daily living, and increased pain. Although the evidence supporting the efficacy of multiple interventions to improve range of motion in neuromuscular diseases in a sustained manner is lacking, there are generally accepted principles with regard to splinting, bracing, stretching, and surgery that help minimize the impact or disability from contractures.

The surgery should only be considered in extreme cases, and must be examined by a multidisciplinary team.[6] A thorough history and physical examination help in an appropriate diagnostic workup, and optimal orthopedic management of each patient.[6]

We inform that, as distal muscle weakness is progressive in these patients, the strengthening of proximal groups appears to be useful for managing gait for as long as possible. Although a group of researchers did not identify overwork weakness after strength and power exercises in patients with CMT, we must be cautious so as not to harm an already ailing system with failed attempts to demyelination and remyelination or even axonal damage.[7]

Treatment of CMT is done in conjunction with medical professionals of various specialties. After diagnosed by a neurologist, CMT patients are usually directed to either a podiatrist for care of their foot problems, an orthotist for the manufacture and fitting of braces, an orthopedic surgeon for interventions to straighten toes, lengthen heel cords or lower arches, and a physical therapist or occupational therapist to design exercise programs to strengthen muscles or learn energy conservation.[3]

An additional problem related to CMT that needs to be addressed by a medical professional is the pain that some patients experience. Pain might be sharp and sudden, or the gnawing, continuous ache of chronic pain. Some pain is associated with dysfunctional nerves that fire sporadically, and some can be attributed to weakened and poorly functioning muscles. Joints and ligaments in the feet and ankles are often painful because of the extra strain put on them by other muscles that have been rendered useless by CMT.

The causes of pain vary, so will the treatments. No one, however, should be told that there is no pain associated with CMT, since pain is experienced in a very personal and individual way.[6]

Lung function tests proved to be minimally abnormal in patients with CMT disease. Therefore, treadmill training and the practice of breathing exercises, besides being safe and well tolerated, induced some post-training positive effects (better exercise tolerance). Strength training or aerobic exercise programs, when well cadenced, might optimise muscle and cardiorespiratory function and prevent disuse atrophy and deconditioning in neuromuscular diseases, such as CMT.[8]

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