

Abstract

Author: Elizabete Pujāte

Title: Case study of a Patient with Charcot-Marie-Tooth disease

Objectives: The thesis is divided into two general parts. The first one consists of an overview of this inherited genetic disease called Charcot-Marie-Tooth, its' discovery, the clinical picture of it, etiology, prevalence in population and currently available treatments. The second part presents the case study, with respective examinations provided, applied therapy and evaluation of results and the therapy effectiveness.

Methods: All examinations and treatments are based on the knowledge I have acquired in the Faculty of Physical Education and Sports of Charles University. These include examination of posture, gait, breathing pattern, goniometry, muscle strength test, muscle length test, anthropometric measurements, neurological examination, fascia examination, scar mobility testing, examination of the muscle tone, joint play examination. The methods used for the patient therapy were mainly strengthening exercises along with DNS and PNF Kabat technique, in addition we performed sensory-motor stimulation and myofascial release, through stretching, soft tissue therapy and PIR. No invasive methods were used.

The main goals of the therapy was to reduce the pain of the patient, improve posture and gait, increase muscle strength mainly in periphery of feet, but also in the trunk core and finally improve quality of activities of daily living (ADL) by preventing any further complications, which would prolong the independence of the patient.

Results: The patient underwent 14 therapy sessions. Due to the patients positive attitude and motivation for improvement, the applied therapy was quite successful. His ADL were improved, as well the main measurable improvements were in posture as the patient now is more aligned according to the perfect standard, gait for he can now walk on his own for 5m and the gait stereotype is better, muscle strength was improved based on grading scale and pain was reduced in cervical spine based on subjective assessment from patient.

Conclusion: The therapies were successful in solving or reducing the limitations and restrictions found during the initial kinesiological examination. The improving of overall condition of the patient was gradual, but by the end of his stay at the hospital both objectively and subjectively there were improvements.

Keywords: Physiotherapy, Charcot-Marie-Tooth disease, peripheral neuropathy