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Effect of intensive and individualized physiotherapy for persons with Chacot-Marie-Tooth disease- a single-subject experimental design study

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European Reference Network for rare or low prevalence complex diseases

/ Introduction

Charcot-Marie-Tooth (CMT) disease is a rare inherited neuromuscular disorder primarily affecting peripheral sensorimotor nerves in the legs and arms. This leads to muscle weakness and sensory deficits, which may also cause structural deformities in the feet. The result is gait and balance difficulties. Although physiotherapy is generally recommended to target these difficulties in individuals with CMT, there is a paucity of studies investigating its effectiveness in the patient group.
The aim of the study was to assess the impact of a two-week intensive and individualized physiotherapy intervention on gait function and balance in adults with CMT.

/ Methods

- **Individualized physiotherapy, 90 minutes daily for 10 treatment days. Focus on integration of postural control in functional tasks, and selective movement as basis for coordinated movement sequences.**
- **Single Subject Experimental Design (SSED), ABA design, 2SD-band method.**
- **Twelve measurements were conducted, four measurements per phase, over a 6 week period.**
  - **Baseline (BL): Week -2 and -1.**
  - **Intervention (IV): Week 0 and 1.**
  - **Follow-up (FU): Week 2 and 3 (A), week 4 (B)**

	Participant A	Participant B
Age/gender	27M	31M
CMT-type:	CMT 1A	CMT 1E
ICD-10:	G60.0	
ORPHA:	101081	90658

Outcome measure	Gait related	Balance related
Zebris© PDM	Stride length (cm) Step width (cm)	CoP average velocity (mm/sek) 95% confidence ellipse sway (mm2)
2MWT	Distance (m)	
Maximum step length (MSL)		Forward, side, backward (cm)
Mini BESTest		Total score
Pain	**Fatigue**	**Perception of change**
NRS 0-10 scale	NRS 0-10 Scale	Patient Global Impression of Change (PGIC)

/ Results

Participant A	
Significant changes (+2SD):	**Non-significant changes (<2SD):**
Step width on PDM (IV and FU)	Stride length, CoP average velocity, 95% confidence ellipse sway on PDM
Mini-BESTest (3 of 4 FU)	2MWT
MSL forward and backward (IV and FU), MSL sideway (FU)	NRS pain
NRS fatigue (IV and FU)	
PGIC *somewhat better* balance (4/7) and *moderate better* walking (5/7)	

Participant B	
Significant changes (+2SD):	**Non-significant changes (<2SD):**
Stride length on PDM (2 consecutive and 3/6 measurements after BL)	Step width, CoP average velocity, 95% confidence ellipse sway on PDM
Mini-BESTest (IV and FU)	2MWT
MSL backward (IV and FU)	MSL forward and sideways
	NRS pain and fatigue
PGIC *much better* balance (7/7) and *better* walking (6/7)	

*** Caution must be exercised in interpreting some of the results due to the variability in measurements at BL. This holds especially for stride length, CoP average velocity and 95% confidence ellipse sway at the PDM and 2MWT for both participants. Participant B also showed variability in NRS pain and fatigue.**
***Participant B underwent 2 of 4 FU-measurements due to unrelated medical complications needing hospitalization.**

/ Conclusion

The findings in this study suggest that intensive and individualized physiotherapy treatment may have a positive impact on objective measures and self-perception of gait and balance functions, as well as fatigue, in adults with CMT.
However, due to the limited sample size, study design, short intervention- and follow-up phase, there is a strong need for additional studies evaluating physiotherapy interventions for individuals with CMT, where these limitations are addressed.