LOWER LIMB JOINT WORK DURING COUNTERMOVEMENT JUMPS IN CHILDREN WITH CHARCOT-MARIE-TOOTH TYPE 1A

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The purpose of this study was to identify lower limb joint power and work in countermovement jumps (CMJ) in children with Charcot-Marie-Tooth type 1A (CMT1A). Therefore, previously collected CMJ data of seven children with CMT1A were compared to seven age- and sex-matched typically developing children (TDC) in terms of the hip, knee and ankle joint angles, moments, power and work. In the knee and ankle joints, significantly lower values of power, positive work, and total work were found in children with CMT1A. Furthermore, lower percentual ankle contributions to total work were found when compared with TDC. This study might be an addition to previous findings concerning impaired jumping mechanics in patients with CMT and aligns with clinical findings, indicating greater impairment of distal muscles.

KEYWORDS: CMT, patients, joint contribution.

INTRODUCTION: Charcot-Marie-Tooth disease (CMT) is one of the most common inherited neurological diseases (Skyre, 1974), with type 1A (CMT1A) being the most frequent. CMT1A is caused by duplication of the short arm of chromosome 17 (Krajewski et al., 2000). Patients show muscle weakness, atrophy, and sensory loss, where the lower limbs are affected more than the upper limbs and the distal muscles more than the proximal muscles, possibly due to a length-dependent degeneration of motor axons (Krajewski et al., 2000). Symptoms such as weakness in the hands, fingers, feet, and legs, difficulties during walking, problems with balance, pain, and others have been reported (Thomas et al., 2022). Furthermore, deformities such as pes cavus can be observed (Maranho & Volpon, 2009). In addition to slowed nerve conduction velocities, compound motor and sensory nerve action potentials are reduced (Krajewski et al., 2000). In CMT1A, the firing rate of motor neurons was lower than that in a healthy population (Noto et al., 2021). This finding is important for the clinical picture, considering that the maximal explosive force generated correlates with the initial recruitment and maximal discharge frequency of motor neurons (Del Vecchio et al., 2019).
To assess lower limb power and strength, vertical jump tests, such as the countermovement jump (CMJ), are often used, and are thus important in many sports (Barker et al., 2018; Cronin & Hansen, 2005). Furthermore, CMJs are used to monitor the neuromuscular status in athletes (Claudino et al., 2017). Several studies have reported the determinants of performance during CMJ. Mcerlain-Naylor et al. (2014) showed that 74% of the performance variation in CMJ height can be explained by CMJ peak knee power, take-off shoulder angle, and CMJ peak ankle power. Other studies have also revealed that the knee is most important for power generation during jumps (Hubley & Wells, 1983), particularly in the eccentric phase, where the knee is the leading joint for work production. In the concentric phase, the hip and knee joints are equally important for generating work (Raffalt et al., 2016). A previous study revealed that children with CMT1A show a reduced jump height due to a reduced net vertical impulse (Alexander & Broser, 2022). The aim of the present study was to compare lower limb joint work during CMJs in children with CMT1A with that in typically developing children (TDC). The hypothesis underlying this study was, in particular, reduced ankle power and work due to the greater influence of CMT on distal rather than proximal muscles.

METHODS: In this retrospective study, data from patients and TDC data from a previous study (Alexander & Broser, 2022) were used and further analyzed. Following, seven children with CMT1A (13.0±1.8 years, 1.57±0.07 m, 41.2±3.9 kg, 4 female; jump height: 13.24±3.49 cm; net

vertical impulse: 321.38±48.27 N/kg*s) were compared to seven sex- and age-matched TDC (12.9±1.7 years, 1.56±0.07 m, 44.9±6.7 kg, 4 female; jump height: 29.06±3.19 cm; net vertical impulse: 456.84±32.76 N/kg*s) were selected. This study was approved by the regional ethics board (BASEC 2021–00314; EKOS 21/025).
After a warm-up involving walking, running, and one to two preparatory jumps, the participants performed three CMJs with each leg on separate force plates (Advanced Mechanical Technology Inc., Watertown, MA, USA; 1000 Hz). Self-reflecting markers were attached according to the Plug-in-Gait model (Kadaba et al., 1990), and data were collected using a ten-camara-based motion capture system (Vicon, Oxford Metrics Ltd., Oxford, UK; 200 Hz). Participants were instructed to squat to their preferred depth, jump as high as possible, and were allowed to use their arms. The decision on when to take the next jump was participant-driven. Trials were repeated in case of landing difficulties or perceived non-maximal effort, assessed through participant self-report. Each participant performed a maximum of five CMJs. Kinematic and kinetic data were filtered using the Woltering method and a 50 Hz Butterworth low pass filter, respectively. The cut-off frequency was defined using residual analysis. Further data analyses were conducted using MATLAB (R2023b; MathWorks Inc., USA). Jump execution was analyzed between the start of the movement (i.e., the time point when the vertical ground reaction force (GRF) subtracted by the body weight was below zero) and take-off, when both feet left the ground. The task was further divided into three phases: the unweighting phase (i.e., negative total GRF impulse), the breaking phase (i.e., braking impulse is equal to the negative impulse), and the net vertical impulse generating phase (i.e., phase in which the net vertical impulse is generated; this is calculated by subtracting the breaking impulse from the positive impulse). The mean of both legs for hip, knee, and ankle sagittal angles and moments, as well as non-dimensional power, were calculated. Furthermore, positive, negative, and total (positive minus negative) work during the net impulse-generating phase at the hip, knee, and ankle joints was computed through the numerical integration of power over time. Additionally, positive, negative, and total joint work (sum of hip, knee, and ankle work), along with each joint's contribution to the total work, were determined. Moment, power, and work data were normalized to body mass. To visualize the entire task execution, ensemble means were computed by time-normalizing angles, moments, and powers.
All statistical analyses were conducted in MATLAB. Differences in CMJ parameters between children with CMT1A and TDC were identified using the statistical parametric mapping (SPM) (Pataky, 2012) equivalent to a t-test for temporal profiles and independent Student's *t*-test for discrete values ($\alpha = 0.05$). Effect sizes were quantified using Cohen's d and categorized as small (d = 0.20–0.49), medium (d = 0.50–0.79), or large (d > 0.80) (Cohen, 1992).

RESULTS: Children with CMT had lower knee and ankle angles, moments, and powers than those with TDC, whereas no difference was observed at the hip (Figure 1). In children with CMT1A, positive and total work were lower across all three joints, being significant for the knee and ankle, while for the hip, although a high effect size was observed, it did not reach the level of significance. Concerning the joint contribution to the total amount of work conducted over all three joints, children with CMT1A had significantly higher negative percentage contributions in all three joints. A medium effect size was observed for positive hip and positive and total ankle joint contribution (Table 1). At the individual joint level, children with CMT1A show significantly higher negative contributions at the knee (CMT: 4.2±1.6%, TDC: 2.3±1.2%, p=0.025, d=1.372) and ankle (CMT: 2.2±1.5%, TDC: 0.7±0.6%, p=0.029, d=1.372). No significant difference was found in the hip joint, although the mean value of CMT patients (7.3±5.1%) was more than twice as high as that in TDC children (3.5±5.1%, p=0.196, d=0.732).

DISCUSSION: Our hypothesis of lower ankle joint power and work due to a greater influence on distal rather than proximal muscles can be accepted. In line with previous studies, lower jump height in CMT1A patients might be associated with lower peak knee (not significant) and ankle power (McErlain-Naylor et al., 2014). Ankle power, as well as knee and ankle work (positive and total), were significantly lower. TDC had the highest contribution to the total joint work at the ankle, followed by the knee and hip, while CMT1A patients tended to have

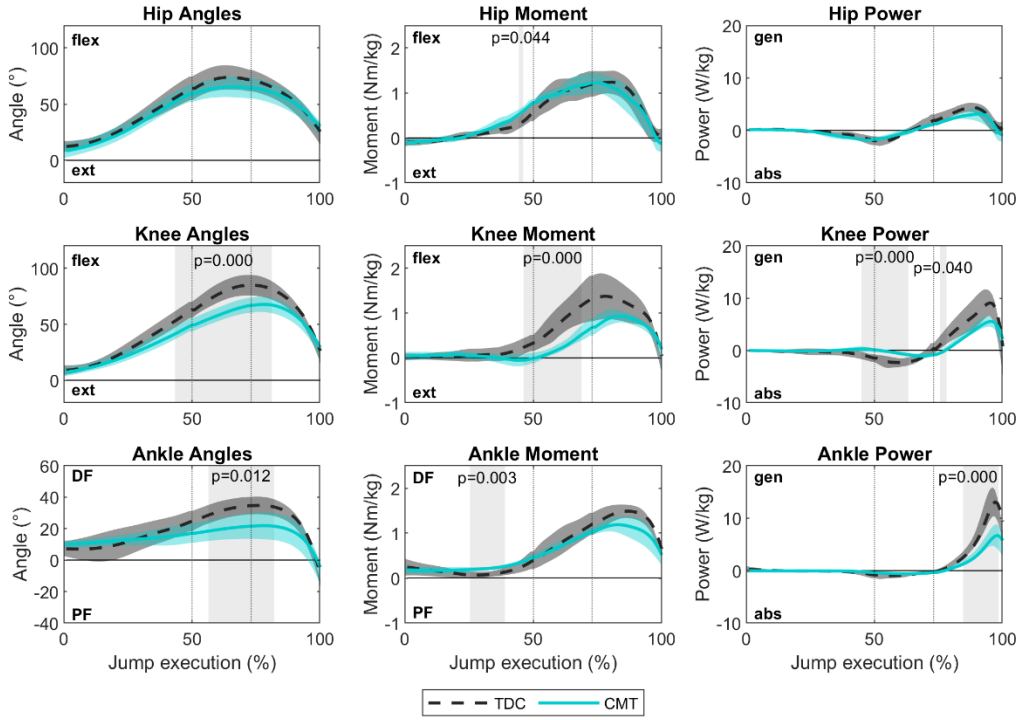

Figure1: Mean and standard deviation for hip, knee, and ankle angles, moments, and powers for children with CMT1A and TDC. The dashed lines indicate the different phases.

Table 1: Mean (SD) joint work of the hip, knee, and ankle joints during the net impulse generating phase of a CMJ, as well as the contribution of the respective joints to the total lower limb work.

	hip			**knee**			**ankle**		
	positive	negative	total	positive	negative	total	positive	negative	total
work (J/kg)									
CMT	0.37 (0.16)	-0.03 (0.02)	0.34 (0.16)	**0.47 (0.1)**	-0.02 (0.01)	**0.45 (0.09)**	**0.46 (0.15)**	-0.01 (0.01)	**0.45 (0.15)**
TDC	0.57 (0.2)	-0.01 (0.01)	0.56 (0.21)	0.92 (0.26)	-0.02 (0.01)	0.9 (0.26)	0.96 (0.15)	-0.01 (0.01)	0.95 (0.15)
p	0.059	0.217	0.053	0.001	0.639	0.001	0.000	0.309	0.000
d	1.117	0.696	1.147	2.270	0.257	2.292	3.321	0.568	3.379
contribution (%)									
CMT	28.6 (5.3)	**-2.2 (1.6)**	26.4 (5.8)	39.1 (5.9)	**-1.6 (0.5)**	37.5 (5.9)	37.0 (4.4)	**-0.9 (0.7)**	36.1 (4.3)
TDC	23.8 (9.6)	-0.6 (0.7)	23.3 (10.1)	37.7 (9.3)	-0.8 (0.4)	37.0 (9.4)	40.1 (5.1)	-0.3 (0.2)	39.8 (5.3)
p	0.272	0.028	0.493	0.736	0.004	0.898	0.245	0.048	0.180
d	0.616	1.338	0.378	0.184	1.919	0.070	0.653	1.174	0.761

Bold values indicate significant differences ($p < 0.05$). p = p-value; d = Cohen's d effect size.

higher hip and lower ankle contributions compared to those with TDC. Although these findings were not significant they align with previous findings indicating greater impairment in distal muscles than proximal muscles in patients with CMT1A (Krajewski et al., 2000). A higher hip joint contribution compared with TDC might be a compensatory mechanism for the weaker distal muscles. Interestingly, the negative contributions of all three joints to the total work, as well as the negative contribution within each joint, were greater for CMT1A patients than for TDC during the net impulse generating phase. Therefore, the task execution of CMT1A patients is less efficient in terms of generating capacity. Furthermore, joint angles, moments

and powers of CTM1A patients compared to TDC might be an indication of a less efficient stretch-shortening cycle during the countermovement.
A limitation of this study was the small number of participants. For further studies a higher number of participants would be desirable also due to the findings of Raffalt et al. (2016) that children have a higher intra-subject variability and an inconsistent movement pattern when compared to adults. Further, longitudinal studies would be of interest to observe changes during aging.

CONCLUSION: Children with CMT1A presented lower hip, knee, and ankle negative and total joint work during the net impulse generating phase compared to TDC. Furthermore, lower ankle joint contributions to the total lower limb work in children with CMT1A compared to TDC align with clinical findings, indicating greater impairment of distal muscles. This study provides a deeper understanding of impaired jump mechanics at a joint-specific level.

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