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Evaluation of physical function before and after medical treatment in non-ambulant patients with spinal muscular atrophy aged 11-25

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Aim

The aim of the study was to assess functional capacity in non-ambulant patients with SMA >10 years of age before medical treatment and six months after start-up, and to learn about their hopes, expectations and experiences regarding medical treatment.

Conclusion

Physical function and perception of fatigue was stable after six month of medical treatment. Although there was a little change in EK2-sum score and FSS score, this was not significant.

In the interview, however, two participants said they had achieved the improvements they had expected and five participants had maintained their functional level. Seven participants had experienced unexpected improvements (table 1).

Results

Fifty-nine people accepted the invitation and took part in the online assessment. Eleven people met the criteria for treatment introduced in July 2023 (≤ 25 years old, and a minimum RULM score of 2); nine of them (4 F, 5 M) aged >10-25 participated in both the baseline and follow-up assessment.

Brooke score ranged from 1-2, RULM score from 2-5 at both assessments. Median EK2 sum-score was 24 at the initial assessment and 23 at the follow-up assessment (p=0.438) (fig 1). Median FSS sum-score was 16 at the initial assessment and 15 at the follow up assessment (p=0.180) (fig. 2).

Table 1 illustrates hopes and expectations before and after medical treatment. All participants expected a positive impact on their quality of life. Five participants hoped for improvement, while only four expected improvements.

After six months of treatment, seven participants experienced improved quality of life, 5/8 had their hopes fulfilled and 7/9 participants found their expectations had been met. The same seven participants furthermore described unexpected improvements in functioning.

Methods

All non-ambulant patients with 5q SMA registered with RCFM > 10 years old (n=117) were invited to participate in a video-recorded online assessment. A questionnaire with open-ended questions about hopes and expectations for the potential medical treatment was included in the invitation.

Assessment of physical function included classification of upper limb function [Brooke et al 1981], hand function by RULM item B (bring hands from lap to table) and item D (pick up tokens/coins) [Mazzone 2016], the EK2 scale [Steffensen et al 2008], and the adapted Fatigue Severity Scale (FSS) with seven items [Werlauff 2014]. The assessment was performed by either an OT or PT.

Those who were offered medical treatment were invited to repeat the assessment six months after initiation of treatment. After the second round of physical assessment, a researcher performed a short interview. The participants were asked about their experiences with medical treatment and whether their hopes and expectations had been met.

For reliability, the prime investigator (UW) reviewed all the recorded assessments to rule out discrepancies; these scores were registered. Differences between first and second assessments were calculated by Wilcoxon signed rank test.

Figure 1: EK-sum score of 17 items at Baseline assessment (EK 1-A-17 SUM) and six months after medical treatment (EK 1-B 17-SUM)

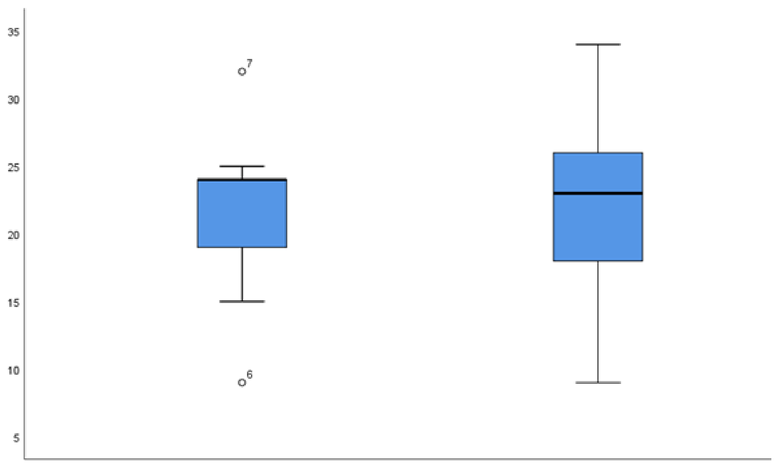
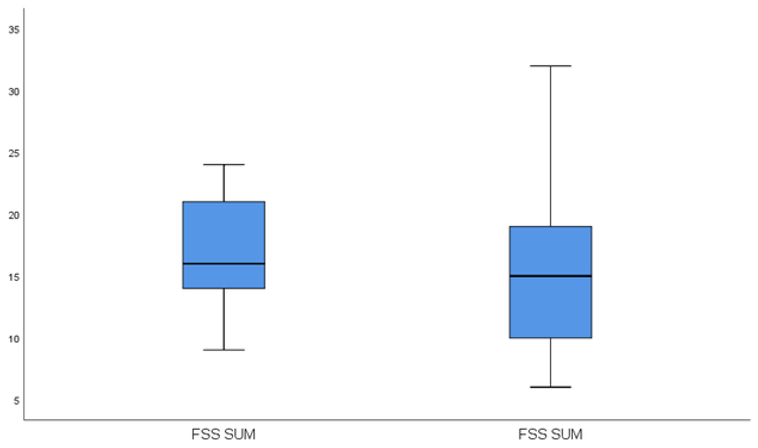
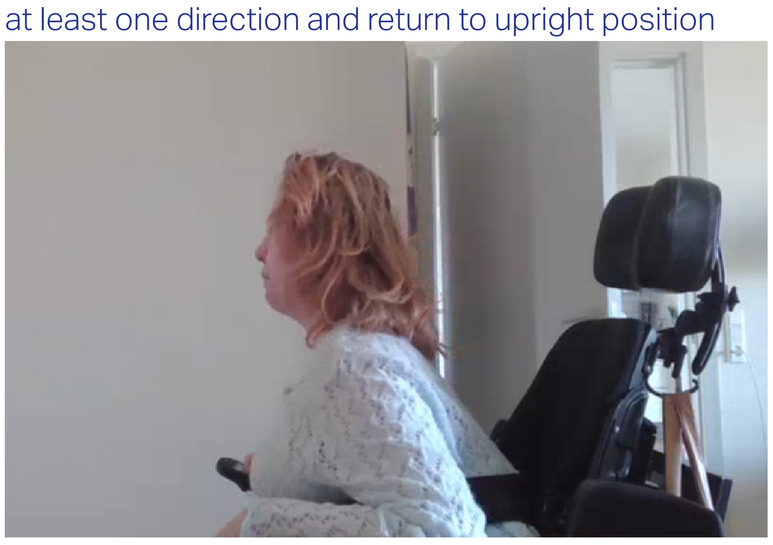


Figure 2: FSS-sum score (7 items) at Baseline assessment and six months after medical treatment



Item EK4. Ability to balance in wheelchair.
Score 1 - Can bend the upper body > 30 degrees in at least one direction and return to upright position



Background

During the spring of 2023, the Danish Medicines Council discussed the possibility of expanding the criteria for medical treatment to patients with 5q spinal muscular atrophy (SMA) which until then had been limited to children up to 10 years of age.

More than 150 people with SMA did not receive medical treatment and were potentially in the new target group for an expansion. Besides a requirement for a minimum level of functioning corresponding to 2 points on the RULM scale for non-ambulant persons, the new criteria for being eligible for treatment were unknown.

We wanted to prepare for a baseline examination, but did not have the capacity to assess all patients physically, so we decided to narrow the target group to non-ambulant patients and assess their functional capacity online. In addition, we wanted to learn about their hopes and expectations for medical treatment and their experiences after medical treatment had been initiated.

Table 1: Hopes and expectations before and with treatment

	HOPES	EXPECTATIONS
BEFORE MEDICAL TREATMENT	Improve functions (n=5) Maintain present functions (n=3) Don't know (n=1)	Improve functions (n=4) Maintain present functions (n=5) Improved QoL (n=9)
SIX MONTHS AFTER START-UP	*HAVE HOPES BEEN FULFILLED?* Improved functioning (n=2/5) Maintained present functions (n=3/3) Hopes not fulfilled (n=4)	*HAVE EXPECTATIONS BEEN MET?* Experienced expected improvements (n=2/4) Maintained level of functioning (n=5/5) Expectations not met (n=2) QoL improved (n=7)
	HOPES FOR THE FUTURE Maintain present level of functioning (n=6) Continued improvement (n=3)	*HAS ANYTHING EXCEEDED EXPECTATIONS?* Better ventilation (n=1) Improved fine motor skills (n=1) More energy (n=3) Feel stronger (n=2)

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