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Feasibility of AI-Based Severity Assessment Using SMCS in Spinal Muscular Atrophy

Jejun Park[1], Jinhee Choi[1], Wonsik Dho[1], Kwangsub Song[2], Sangui Choi[2], So Ra Moon[2], Hooman Lee[2], Jin Hong Shin[3], Howon Jeon[4], Soo-Yeon Kim[1,4,5*]

Department of Rehabilitation Medicine, Pusan National University Yangsan Hospital[1]
Department of AI Research, EXOSYSTEMS[2]
Department of Neurology, Pusan National University Yangsan Hospital, Pusan National University School of Medicine[3]
Department of Rehabilitation Medicine, Pusan National University School of Medicine[4]
Research Institute for Convergence of Biomedical Science and Technology, Pusan National University Yangsan Hospital[5]

Introduction

Spinal muscular atrophy (SMA) is the most prevalent motor neuron disease in children. Current assessments, including the Hammersmith functional motor scale expanded (HFMSE) and manual muscle test (MMT), rely on subjective evaluation and frequent hospital visits. This study evaluated the feasibility of an artificial intelligence (AI) based severity assessment technique using stimulated muscle contraction signals (SMCS) to reflect functional motor performance and muscle strength.

Methods

This cross sectional study was conducted from March 2023 to December 2024 and enrolled genetically confirmed SMA patients, excluding those with intellectual disabilities or neurological deficits other than SMA. SMA severity was defined using an integrated index combining the HFMSE and normalized objective muscle strength (ankle, knee, wrist, and elbow). SMCS were collected from 6 body parts using a wearable surface electromyography (sEMG) electrical stimulation device at 5–30 Hz and analyzed after frequency-wise segmentation and fast Fourier transform (FFT) based feature extraction. Features with an absolute correlation coefficient >0.25 were selected, and an artificial neural network (ANN) regression model with leave one out cross validation (LOOCV) was used to estimate SMA severity (Fig. 1).

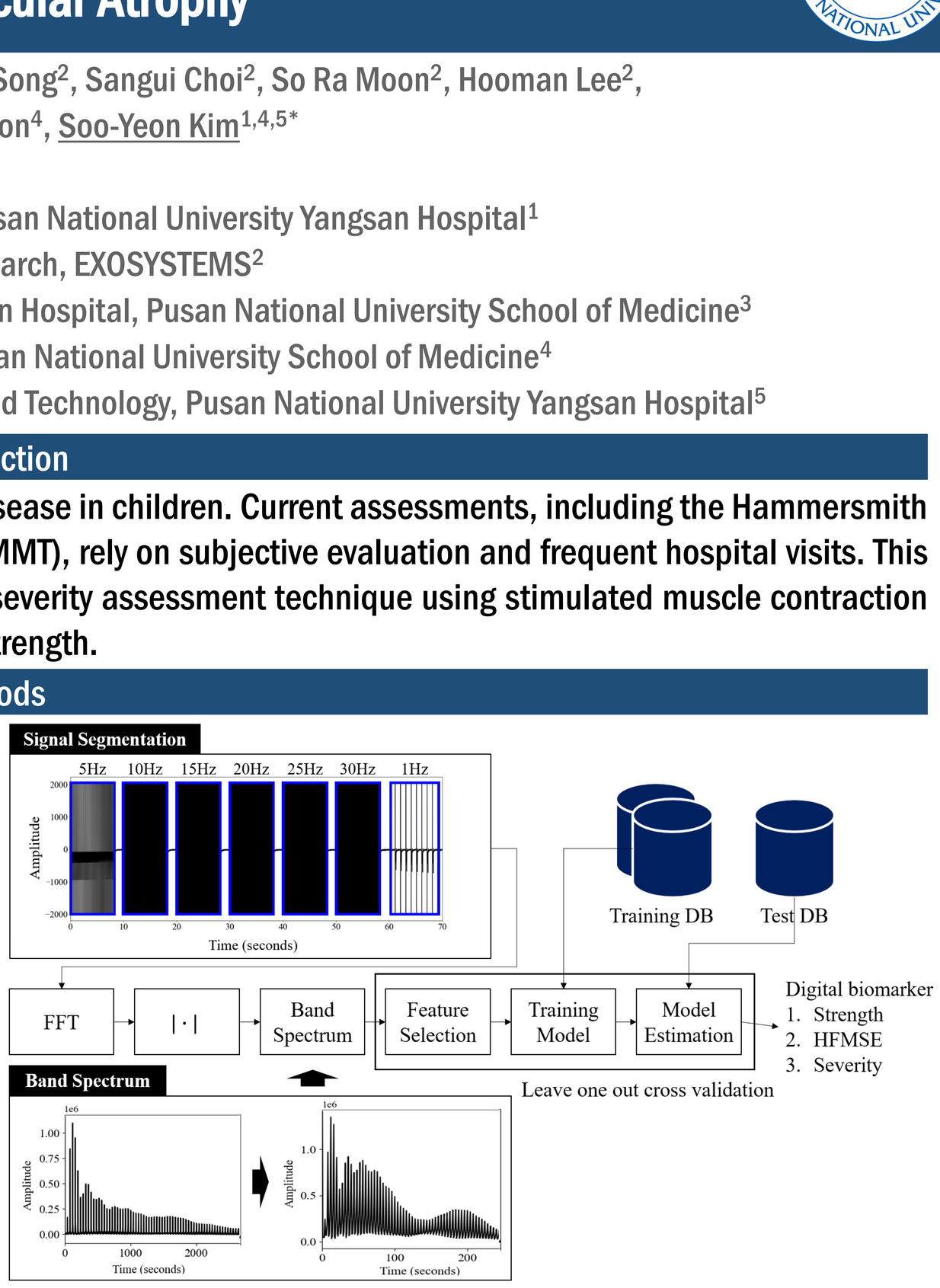

Figure 1. Total block diagram

Results

A total of 21 patients with SMA were analyzed (type 2, n=14; type 3, n=7). There were no significant sex differences in age, height, body weight, or HFMSE score (all p>0.05). Model performance for the functional motor assessment index, muscle strength index, and severity digital biomarker were summarized (Table 1). The ANN models showed reasonable predictive performance for the functional motor assessment index (r=0.74) and muscle strength index (r=0.70) (both p<0.001). For the severity digital biomarker, the model showed r= 0.76, standard deviation of error = 3.35, and mean error = 0.00 (p<0.001). Scatter plots showed significant correlations between model estimations and actual values for all three outcomes. Bland-Altman plots showed uniformly distributed errors across the measurement range, suggesting low bias and balanced model performance, and supporting the validity of the proposed approach (Fig. 2).

Table 1. Model estimation for functional motor assessment, muscle strength, and digital biomarker

Functional motor assessment index				Muscle strength index				Severity digital biomarker			
ME	STDE	r	p-value	ME	STDE	r	p-value	ME	STDE	r	p-value
0.03	0.20	0.74	<0.001*	0.66	1.10	0.70	<0.001*	0.00	3.35	0.76	<0.001*

ME; mean error, STDE; standard deviation of error, Asterisk means statistically significant (p<0.05).

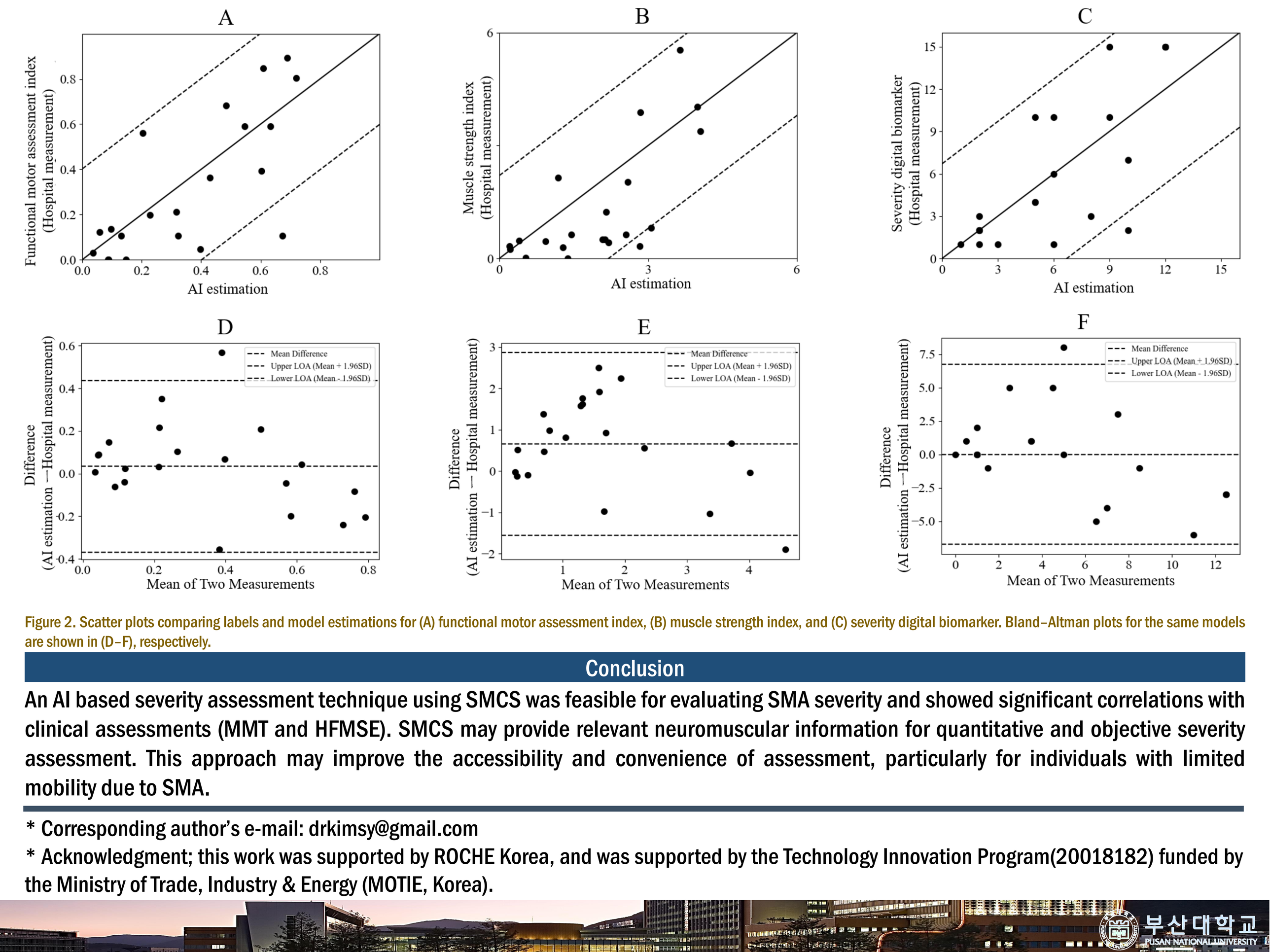

Figure 2. Scatter plots comparing labels and model estimations for (A) functional motor assessment index, (B) muscle strength index, and (C) severity digital biomarker. Bland–Altman plots for the same models are shown in (D–F), respectively.

Conclusion

An AI based severity assessment technique using SMCS was feasible for evaluating SMA severity and showed significant correlations with clinical assessments (MMT and HFMSE). SMCS may provide relevant neuromuscular information for quantitative and objective severity assessment. This approach may improve the accessibility and convenience of assessment, particularly for individuals with limited mobility due to SMA.

* Corresponding author's e-mail: drkimsy@gmail.com

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