Single Breath Count Test in Remote and In-Person Monitoring of Respiratory Function in LOPD and NMDs – a single center study

Ignazio Giuseppe Arena[1], Mattia Porcino[2], Maria Chiara Bannino[2], Cristian Usbergo[2], Antonio Toscano[2], Musumeci Olimpia[2]

[1] BIOMORF Department, University of Messina; [2] Department of Clinical and Experimental Medicine, University of Messina

Introduction: Respiratory involvement is a frequent and clinically significant feature in Neuromuscular diseases, including late-onset Pompe disease (LOPD). The Single Breath Count Test (SBCT) is a simple, non-invasive tool for assessing respiratory endurance, previously explored in NMDs such as myasthenia gravis, including some applications in remote settings. However, data on its feasibility and validity when performed remotely either on certain specific neuromuscular conditions are currently lacking.

Objective: This study aimed to evaluate the feasibility, reliability, and clinical utility of the SBCT in patients with NMDs, with a particular focus on LOPD. We compared SBCT results to standard spirometry and assessed its potential as a practical outcome measure.

Methods: A total of 28 patients with genetically confirmed LOPD, 15 patients with other NMDs, and 15 healthy controls underwent SBCT and spirometry in both upright and supine positions, during both in-person and remote assessments (Figure 1). Additional evaluations included the 6-Minute Walk Test (6MWT), Fatigue Severity Scale (FSS), and SF-36 quality-of-life questionnaire, maximal inspiratory and expiratory pressure (MIP and MEP). Data were adjusted for age and sex and analyzed using Pearson correlation, paired t-tests, and ANOVA.

Results: SBCT scores were significantly lower in LOPD and NMDs patients compared to healthy controls ($p < 0.05$), with good discriminatory ability when considered on FVC values (Figure 2). Age influenced both SBCT and spirometry outcomes, while no sex-related differences were observed. A mild but consistent correlation was found between SBCT and spirometry, both in-person and remotely, particularly with percent-predicted FVC ($p < 0.05$; Figure 3 and 4). Importantly, SBCT showed good reproducibility across the two different assessment settings and might correlate with the need of NIV.

Figure 1. Distribution of the SBCT score in patients and controls

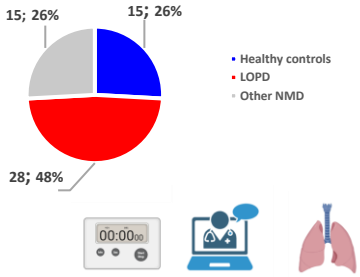


Figure 2. Distribution of best SBCT scores among different groups

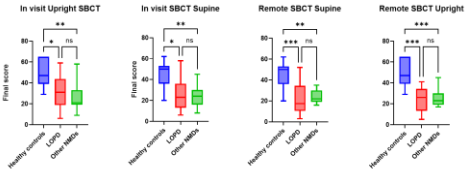
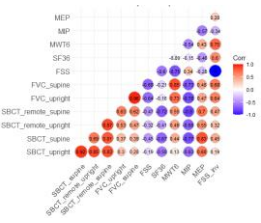


Figure 3. SBCT correlation with other functional measurement in LOPD



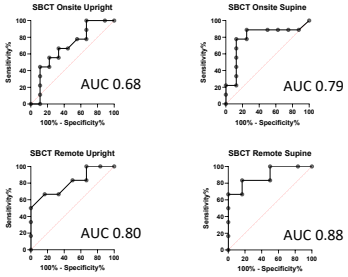
SBCT (supine) – MEP; r= 0.83; Better expiratory strenght -> improved SBCT performance

SBCT – FSS (inverse); r=0.45; Less perceived fatigue -> better SBCT scores

SBCT –MIP; R= -0.77; Poor inspiratory strenght associated with lower SBCT

SBCT – SF36; R = -0.58; Lower quality of life scores relate to poorer SBCT performance

Figure 4. Can SBCT scores predict NIV?



Conclusion: These findings support the SBCT as a feasible, low-burden method for detecting respiratory impairment in LOPD, even in early stages of disease. Its simplicity, non-invasiveness, and reliability - especially in remote contexts - highlight its potential as a valuable additional instrument to conventional pulmonary testing in both routinary and clinical trial settings, particularly where access to standard assessments is limited.