

Neuropsychological Profiles, Sleep Disorders, and Behavioral Adaptation in Smith-Magenis Syndrome: Clinical Correlates and Rehabilitation Perspectives

Friday, September 12, 2025 5:30 PM (10 minutes)

Objective: This study aims to identify neurobiological correlations between cognitive, emotional/behavioral, and adaptive functioning, sleep disorders, and specific neuropsychological domains in individuals with Smith-Magenis Syndrome (SMS). A secondary aim is to define more effective rehabilitation strategies targeting deficient domains to support functional and adaptive improvement.

Participants and Methods: The study included 45 individuals with SMS (ages 0–33). Two assessments were conducted: the first examined cognitive, adaptive, and emotional/behavioral development; the second explored sleep disorders and neuropsychological areas such as attention and visuomotor integration. Instruments used included WISC-IV, WAIS-IV, CBCL, Vineland-II, Bells Test, VMI, SDSC, and the Self-Injurious Behavior Scale (SRCA).

Results: Cognitive assessments showed global impairments, with significantly below-average IQ scores (FIQ, VCI, VSI, WMI, PSI). Adaptive functioning was also notably reduced. A strong correlation was found between IQ and adaptive behavior ($p < 0.001$). Emotional and behavioral results showed borderline or low scores, especially on Externalizing and Total Problems Scales. Self-directed aggressive behaviors correlated with internalizing traits ($p < 0.001$), affecting adaptive outcomes ($p < .05$). Neuropsychological testing revealed marked deficits in selective/sustained attention ($p < 0.001$) and visuomotor integration ($p < .01$). Sleep disturbances—mainly DIMS and SWTD—were frequent. Sleep Breathing Disorders correlated with externalizing symptoms ($p < 0.001$); excessive sleepiness was linked to internalizing traits ($p < .01$), more evident in males ($p < .05$).

Conclusion: The data suggest neurophysiological links between sleep regulation and neurobehavioral functioning in SMS, potentially influenced by genetic and epigenetic mechanisms. Identifying these correlations is key to developing targeted, effective rehabilitation strategies.

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Primary author: Dr LIBERATI, Luca (Clinical Psychological Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy)

Co-authors: Dr ARCANGELI, Valentina (Clinical Psychological Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy.); Dr MARFOLI, Angelica (Clinical Psychological Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy.); Dr VANNUCCINI, Sofia (Clinical Psychological Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy.); Dr MARTELLI, Brenno (Clinical Psychological Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy.); Dr GUIDI, Eleonora (Clinical Psychological Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy.); Dr VEREDICE, Chiara (Pediatric Neuropsychiatry Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy.); Dr ONESIMO, Roberta (Rare Diseases Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy.); Prof. CHIEFFO, Daniela Pia Rosaria (Clinical Psychological Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy.); Department Women Children and Public Health, Catholic University of Sacred Heart, 00168 Rome, Italy.)

Presenter: Dr LIBERATI, Luca (Clinical Psychological Unit, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, 00168 Rome, Italy)

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