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Behavioral Interventions for Enhancing Adaptive Skills of Children with Angelman Syndrome

Editorial

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Abbreviations: AS: Angelman Syndrome; AT: Assistive Technology; MBP: Microswitch-Based Programs.

Angelman syndrome (AS) is a rare genetic disorder, first described by Harry Angelman in 1965 [1]. Thus, three of his patients showed severe to profound intellectual disabilities, excessive laughing, jerky movements, and abnormal physical development. Because the three participants emphasized a flat head disorder, they were called "puppet children". Their common behavior was characterized by ataxia, lack of speech, learning difficulties, seizures, tongue protrusion, and motor impairments. A new unique distinct syndrome known as AS was identified.

Individuals with AS typically exhibit hyperactivity disorders and restless behaviors, hypotonia, microcephaly, wide gait, abnormalities in EEG patterns, widely spaced teeth, hypo-pigmentation (i.e., blond hair and light eyes), prominent chin and deep set eyes. Beside the severe to profound range of intellectual delays, autism spectrum disorders are frequently included. That is, lack of communication, reduced social interactions, and stereotyped movements are recognizable [2, 3]. The genetic cause of AS is due to the deletion in the long arm of the 15th chromosome [4]. Furthermore, the E6-AP (i.e., E6 protein encoded by the UBE3A gene) was found to be responsible for AS [5].

In light of the above, one may argue that children with AS may be quite passive and isolated towards their environment with negative outcomes on their quality of life. In fact, their clinical conditions may seriously hamper their social desirability, image, and status. One way to overcome this issue is to envisage assistive technology-based interventions (AT), cognitive-behavioral strategies, and/or behavioral approaches. Accordingly, their self-determination and independence should be fostered, with beneficial effects on both caregivers and families burden reduction [6]. Nevertheless, if AT and AS were inserted in SCOPUS database as keywords, no records were found [7]. Switching on cognitive-behavioral interventions and AS, only two documents were extracted [8, 9]. Finally, searching with behavioral interventions and AS, four reviews and twelve articles were detected in the last five years [10-24].

For example, Tones et al., [14] assessed the protocol of the Global AS Registry and some initial findings. Due to the rare disease and the variance of its phenotype, the registry team strived for case ascertainment completion. Both parents and caregivers submitted data to the registry through a secure internet connection. The registry consisted of 10 modules that covered patients information on (a) demography, (b) history, (c) behavior and development, (d) medications, (e) interventions, and (f) sleep. The results showed that the majority of the registered participants (i.e., four hundred and seventy) were children (i.e., only the 16% were aged over 20 years), most of them (i.e., 76%) reported a chromosome deletion, with a minority reporting a mutation or an imprinting defect (i.e., 20%).

Trickett, Heald, and Oliver [17] conducted semi-structured interviews with parents of fifty children with AS, who experienced sleep problems. The relevance of considering parental experience was emphasized by the variance in the coping process. Thus, 20% of the parents did not need any additional support. Among those who claimed for a further support, the 27% requested a behavio-

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ral intervention and supplementary information about the trajectory of sleep problems in AS (i.e., 18%). The priority for behavioral interventions aimed at supporting both parents and children with AS in improving sleep quality and well-being was evinced, beside longitudinal research into sleep disturbances.

Heald, Allen, Villa, and Oliver [18] exposed four children with AS to a multiple arrangement focused on an alternated social reinforcement and extinction-based behavioral program, cued using a novel stimulus. Twenty-five to thirty-five discrimination sessions were collected and levels of approach behaviors were monitored before and after discrimination trials for two children. All the participants demonstrated the capacity of discrimination after 16-20 sessions as recorded by lower rates of social approach behavior during the extinction process. Reversal effects were evident for the two children exposed to such experimental condition. Results outlined that after a repeated training, the use of a novel stimulus may be helpful as a cue for children with AS to discriminate the adult availability. However, a strong need for sustained teaching procedures within this population was warranted.

The concise overview above reported pointed out the urgency for new further empirical contributions in the literature for both research and practice. For instance, one may envisage AT-based interventions aimed at promoting adaptive skills and simultaneously reducing challenging behaviors of children with AS. Thus, depending their level of functioning, both economical and human resources, and rehabilitative objectives, one may design different programs. Basic forms of AT are represented by microswitches (i.e., electronic tools enabling individuals with multiple disabilities with the independent access to positive stimulation). Although no specific rules exist, some useful guidelines may be detailed for a successful implementation of a microswitch-based program (MBP), based upon learning principles (i.e., causal association between a minimal behavioral response and environmental consequences). First, a plausible response should be identified (i.e., the response should be already available in the individual repertoire, easily produced without excessive effort, it should be directly measurable, and exhibited with verbal or physical prompts). Second, a suitable microswitch should be adopted. That is, the electronic tool should be reliable and be able to detect the behavioral response correctly. Third, a rewarding positive stimulation should be screened. In fact, the positive stimulation should be adequately and sufficiently compensate the response cost. Whenever the three basic guidelines were observed, the MBP would be useful and helpful for the learning process [25].

For individuals with a very low behavioral repertoire and severe to profound intellectual disabilities, one may plan an independent access to positive stimulation [26]. For children with AS who are estimated with developmental disabilities and moderate to profound intellectual delays, one may argue on a combined program of microswitch and VOCA, with the opportunity of requesting the social contact with parents or caregivers, or the possibility of choice between both options [27]. Furthermore, one may envisage a MBP with contingent positive stimulation focused on supporting the locomotion fluency [28]. Finally, a technological intervention based on request and choice alternatives may be implemented [29].

In light of the above, new research within this framework should deal with the following topics. First, microswitch-cluster technology pursuing the dual goal of increasing an adaptive responding and reducing a challenging behavior (e.g., manipulation objects and tongue protrusion) could be arranged. Second, academic performance and on-task behaviors through computer and MBP should be enhanced. Third, the effects of such interventions on participants' indices of happiness and/or positive participation as outcome measure of their quality of life should be recorded. Fourth, maintenance/generalization, post-intervention checks, and/or follow-up phases and sessions to assessing the learning consolidation should be collected. Fifth, participants' preference checks and/or social validation procedures involving external raters to verifying the clinical and social intervention validity should be carried out.

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