An Overview of Cognitive-Behavioral Interventions for Promoting Adaptive Skills of Children with Angelman Syndrome

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Abstract

This paper provides the reader with a concise overview of the empirical evidences available in the last decade on the use of cognitive-behavioral interventions for promoting adaptive skills of children with Angelman syndrome. The results were fairly positive, although no documents were found on the implementation of assistive technology-based programs. Some useful suggestions for future research and practice within this framework were briefly discussed.

Angelman syndrome (AS) is a rare genetic disorder due to abnormalities, mutations, and/or deletions of the ubiquitin-protein ligase (i.e., UBE3A) enzyme encoded by the UBE3A gene, located on the 15th chromosome, first described by Angelman [1]. Based on the observations of three “puppet” children, who presented stiff and jerky gait, lack of speech, seizures, excessive laughing, a new pattern of symptoms was detected. Subsequently to an apparent regular birth, between 6 and 12 months developmental delays emerge. Microcephaly, tongue protrusion, sleep disturbances, aggression, ataxia, intellectual disabilities, communication impairments, hypotonia, unsteadiness, and learning difficulties, feeding problems are commonly included. The diagnosis can usually be made between 1 and 4 years from DNA results (i.e., at least for the 80% of the cases). Hence, AS children may be considered individuals with severe to profound developmental disabilities [2,3]. Accordingly, rehabilitative interventions are undoubtedly mandatory. Beside pharmacological treatments, traditional physiotherapy, and stimulation sessions, one may envisage cognitive-behavioral programs, focused on (a) enhancing adaptive skills, and (b) reducing challenging behaviors, which may interfere in their daily life and/or be deleterious for their or other safety and/or seriously hamper their social image, desirability and status [4].

A computerized search in SCOPUS of the literature available in the last decade (i.e., 2005-2016) revealed 19 documents found by using “Angelman syndrome and behavioral interventions” as keywords, with 5 review papers selected [5-9]. For example, Heald, Villa, and Oliver [10] exposed four children aged between 5 and 10 years to a multiple schedule arrangement (i.e., social reinforcement and extinction were systematically alternated) to evaluate whether high rates of social approach behavior could be modified. Notably, 25-30 discrimination trials (i.e., training sessions) were conducted through a triple AB experimental sequence for all participants. Pre and post-test measures of social approach behaviors (i.e., social attention request) were recorded for two children. All participants emphasized awareness of the distinction between both social reinforcement and extinction conditions after 16-20 intervention sessions. Results indicated the effectiveness of a new stimulus to serve as discrimination for adult availability, through a repeated training.

Summers [11] assessed the impact of approaches based on principles of the applied behavior analysis (ABA) on neuro-developmental outcomes in children with AS. Specifically, a non-randomized pre-test post-test control group design was implemented. The intervention group consisted of 4 AS children aged between 3.1 and 9.2 years. Controls included other AS children who were individually matched on the basis of their chronological age, gender and molecular sub-type. Children recruited for the intervention group were provided with 2-3 ABA-based therapy sessions per week, over a 1-year intervention period. Standard outcome measures of cognitive, adaptive and language skills were recorded at the initial baseline and after 1 year. Although no statistical differences between both groups were pointed out neither at baseline nor after 1 intervention year, positive trends...
were recorded for the intervention group on both cognitive and adaptive skills.

Switching on Angelman syndrome and challenging behaviors domain (i.e., by using the combined keywords in SCOPUS), 8 documents were found, with 1 review paper selected [4], within the above reported time range interval (i.e., 2005-2016). For instance, Strachan, et al. [12] carried out a functional assessment with 12 AS children (i.e., age range 5.0-9.9 years) to investigate the role of aggressive behavior for the recruited participants. Furthermore, the study examined the influence of social contact on smiling, laughing, and social approach behavior in AS children. Levels of aggressive behaviors were experimentally manipulated through different conditions of adult attention and demand. Crying, frowning, laughing, physical initiation with an adult and smiling were additionally recorded. The aggressive behavior was observed among 10 participants. One participant demonstrated that the challenging behavior (i.e., aggression) was maintained by attention, social interaction was the main function of aggression for three children, and demand escape explained the aggressive behavior for two children. Although the expected social attention function of the challenging behavior was not confirmed by the data, results suggested that a combined condition of social contact and positive affect was responsible for aggression to serve to both maintain and initiate social interaction. Negative results in terms of unexpected outcomes may have been influenced by the age of the participants and low levels of aggressive behavior observed. Radstaake, et al. [13] used both functional assessment and functional communication training with 3 AS children who exhibited challenging behavior. Both protocols were implemented during classroom and administered by their teachers. Two basic functions for the challenging behavior were identified, namely (a) demand escape, and (b) tangible items. Physical contact with the teacher was emphasized as precursor of the challenging behavior for one child. All participants significantly decreased their levels of challenging behavior once exposed to the functional communication training intervention. Thus, functional equivalence of both challenging and replacement behaviors was empirically demonstrated.

Finally, by inserting “Angelman syndrome and communication interventions” as keywords, 10 documents were evinced, with 1 review paper [14]. Furthermore, two documents were excluded as they were not English written. Within this framework, Hyppa Martin, Reichle, Dimian, and Chen [15] proposed vocal, gestural, and graphic communication modes to a toddler with AS to identify the most efficient way to learn communication skills by the participant. Symbols representing preferred items were introduced following vocal, graphical and gestural modalities according to an alternating treatments single-subject design. Conventionally accepted prompting strategies were adopted to teach symbols in each communication mode. Because the participant did not vocally imitate, vocal mode started with increasing vocal frequencies as first step. Results showed that the graphic mode produced higher levels of communication performance. Calculator [16] describes communication patterns and alternative and augmentative communication programs for AS individuals through parents self-administrated and web-based surveys using Qualtrics software. A series of rating scales and open questions were addressed to acquire information about individuals’ current methods of communication. Outcomes measures of associated importance, usefulness, success, acceptance, and functionality were carried out. Results revealed that alternative and augmentative communication-aided strategies were the most efficiently used, with a specific interest for i-Pad. Differences were detected once ages and education were considered as independent variables. Mobile technologies were strongly associated with beneficial effects.

Surprisingly with the above, if “Angelman syndrome and assistive technology” were used as keywords in SCOPUS, no documents were found. Assistive technology (AT) refers to any electronic device, equipment, piece, or tool enabling persons with severe to profound developmental disabilities and extensive motor impairments with self-determination and independence towards the outside world [17-19]. An AT-based program is built on learning principles (i.e., causal association between a behavioral response and environmental consequences) ensuring an individual with severe to profound intellectual deficits and/or motor inabilities to positively interact with the surrounding items, by promoting constructive engagement [20,21]. Depending upon AS individual’s levels of functioning, both financial and human resources, the rehabilitative goals, and the targeted behaviors, one may envisage different intervention strategies [22,23]. For children with very low behavioral repertoire, one may design a simple micro switch-based program (i.e., AT basic tool connecting a minimal response to positive stimulation through a system control unit) aimed at improving adaptive responding for the independent access to preferred items [24]. Else, systematic comparisons between two or more forms of alternative and augmentative communication may be considered (e.g., PECS and VOCA) [25]. Otherwise, aided programs focused on promoting both ambulation responses and locomotion fluency may be examined [26,27]. Additionally, cluster technology for the simultaneous dual goal of enhancing an adaptive behavior and reducing a challenging responding could be implemented [28]. Moreover, computerized programs for request and choice of preferred items may be applied [29,30]. Finally, occupation strategies may be implemented [31,32]. Indices of happiness and/or positive participation could be recorded as outcomes measures of the participant’s quality of life and social validation assessments involving external expert raters (e.g., students, parents, teachers, caregivers) could be carried out for corroborating the intervention clinical validity [33,34].
Concluding, in light of the above, although the results and the outcomes of the concise selective overview on cognitive-behavioral interventions for AS population were fairly positive, promising and encouraging, new research efforts within AT-based programs for AS children are surely warranted and should deal with the following topics: (a) finding out new individualized suitable and effective technological solutions, (b) assessing the effects of such interventions on indices of happiness and/or participation of the participants, (c) carrying out social validation assessments, and (d) proposing preference checks finalized at investigating participants supports (i.e., agreements).

References


