UNDERSTANDING COMMUNICATION METHODS FOR CHILDREN WITH ANGELMAN SYNDROME: A REVIEW OF LITERATURE AND RESEARCH STUDY PROPOSAL

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ABSTRACT

Angelman Syndrome is a complex genetic neurodevelopmental disorder that is caused by genetic abnormalities. In almost all cases, it causes severe learning difficulties, seizure disorders, motor impairments, ataxia, a happy, sociable disposition, hypopigmented skin and light hair and eye color. People with Angelman Syndrome often have no speech acquisition. If speech is achieved, it is limited to a maximum of about 10 words in total, but most cases only proficiently use about 2-3 words utterances. This is a common struggle and causes distress for both the child and their family. But with the introduction of augmentative and alternative communication devices, it is proposed that communication can be achieved in children with Angelman Syndrome. This project proposes a study and intervention method in which these communication devices can be used to initiate communication among children with Angelman Syndrome.

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INTRODUCTION

There are a small number of research studies on Angelman Syndrome in general, and specifically the area of functional communication skills among individuals with Angelman Syndrome is underdeveloped. The goal of this research paper is to review existing research on Angelman Syndrome in support of a rationale for an informed hypothesis and proposed single case design study to develop a system for acquiring functional communication skills in children with Angelman Syndrome.

WHAT IS ANGELMAN SYNDROME?

Intellectual disabilities (ID) are categorized, usually upon diagnosis and assessments, by mild, moderate, severe, and profound depending on the level of dependency and language capability. About two percent of the population have the diagnostic criteria to meet these categories. People with mild ID often live on their own in residential homes, with family, or with direct care professionals and can receive assistance in order to find jobs and financial security, (Gentile, 2019). People with moderate ID will need varying levels of support from their families as well as community agencies. Their expressive language skills are usually more limited so they can have a more difficult time trying to communicate complaints about their mental health and medical needs, (Gentile, 2019). People with severe and profound ID often are dependent on external supports systems and usually have associated medical conditions, with most individuals requiring assistance for all aspects of life. Significant medical complications such as, "seizure disorders, swallowing difficulties, speech impairments, ambulation limitations, sensory deficits, and reduced life expectancies, are more common for persons in the profound impairment category," (Gentile, 2019, p.2). In most cases, people with Angelman Syndrome will fall within the severe to profound category.

Angelman Syndrome is a complex genetic neurodevelopmental disorder that is caused by genetic abnormalities of the 15q11-13 chromosomal region. These abnormalities include a deletion from the maternal gene, imprinting defects, and mutations within the UBE3A gene. The genetic mechanisms all interfere with this gene's expression, as well as 85%-90% of those also included the clinical phenotypes, (Clayton-Smith & Laan 2003). These phenotypes include subtle facial features such as a wide smiling mouth, prominent chin, microcephaly, and deep-set eyes. People with Angelman Syndrome have a behavioral phenotype as well, including, "jerky movements, frequent and sometimes inappropriate laughter, a love of water, and sleep disorder," (Clayton-Smith & Laan 2003). In all cases of gene deletion, this disorder includes severe learning difficulties, a seizure disorder with a characteristic EEG, motor impairments, ataxia, a happy, sociable disposition, hypopigmented skin, and light hair and eye color, (Clayton-Smith & Laan 2003). Most children with this disorder are diagnosed after presenting with a significant delay in developmental milestones and a slowing of head growth during the first year of life, and speech does not usually develop, but if it does it is extremely limited, (Clayton-Smith & Laan 2003). After an analysis of these various phenotypes, genetic testing of the 15q11-13 chromosomal region is done in order to confirm the preliminary diagnosis of Angelman Syndrome. This, of course, is a much-decreased list of the characteristics and diagnostic processes associated with Angelman Syndrome that has been shortened for understanding.

DIFFICULTIES IN COMMUNICATION

There are few studies that have been able to successfully document the functions of communication skills of individuals with Angelman Syndrome. This is partially in part because it is a relatively "new" disorder, due to diagnosis by genetic testing that has only become available in the past thirty years. In many cases, Angelman Syndrome is preemptively misdiagnosed as

Cerebral Palsy in conjunction with Autism Spectrum Disorder before any genetic testing is done, (Angelman Syndrome Foundation). This greatly affects the way in which proper intervention techniques are implemented. As previously stated, people with Angelman Syndrome often have no speech acquisition. If speech is achieved, it is limited to a maximum of about 10 words in total, but most cases only proficiently use about 2-3 words utterances, (Didden, et. al., 2009). In terms of social cues typical of the United States and other western countries, people with Angelman Syndrome usually do not have problems with social conventions such as making eye contact, laughing, receiving touch as well as touching others, pointing, crying, etc., (Didden, et. al., 2009). Though studies have also found that people with Angelman Syndrome consistently struggle with vocalizing their wants and requests, and properly utilizing American Sign Language, and pointing at objects. Although, these same studies found that about 30% of the individuals with Angelman Syndrome in their samples utilize some form on symbolic, non-speech, form of communication including picture exchange systems, (Didden, et. al., 2009). This suggests that there may be a functioning relationship in implementing alternative and augmentative forms of communication in order to achieve functional communication skills in people with Angelman Syndrome.

PURPOSE

The purpose of this capstone project is to review and analyze the limited amount of research on Angelman Syndrome, as well as propose a single case design study by describing the procedures, including methods of instruction, to further investigate ways to improve functional communication skills in children with Angelman Syndrome. Given that augmentative and alternative communication seems to be promising for individuals with Angelman Syndrome, there is a need for intervention research in order to validate instructional procedures for use with

individuals with Angelman Syndrome (Duker et al. 2004). At present, there appear to be no such studies with appropriate experimental controls to guide communication intervention for individuals with Angelman Syndrome. In order to propose a research study that is built upon previous research, a literature review has been conducted.

REVIEW OF LITERATURE

COMMUNICATION

Most people with Angelman Syndrome do not develop speech and will only have a vocabulary of two to three words. Although, most will be able to understand simple commands that are within their daily routine. Some can communicate with American Sign Language or a picture exchange system, while others use gestures to communicate, (Clayton-Smith, 2003). Most people with Angelman Syndrome understand many single words, including the names of familiar objects, familiar people, as well as simple action words, but understanding small phrases that focus on concrete objects are more easily understood than those focusing on concepts or abstract relationships, (Dan, 2008). Expressive verbal communication is generally less developed than receptive communication. This discrepancy often leads to frustration and can decrease the quality of communication, (Dan, 2008). Oral sounds are rare, and oral dysfunction has not been studied in Angelman Syndrome. Though when sounds do occur, they are limited to vowels or a few consonants, and this may suggest deficits in cerebellar dysfunction as well as deficits in various motor systems, (Dan, 2008). It is apparent that nearly all people with Angelman Syndrome, especially as children, demonstrate an openness towards social interactions and often initiate communication. Children with Angelman Syndrome generally do not have a deficit in theory of mind [which is the ability to attribute mental states to ourselves and others and allows the ability to predict and interpret the behaviors of others] and do not have social apprehension,

(Dan, 2008). Because of this, children with Angelman Syndrome can utilize the information in social situations in order to learn the meanings of words.

COMMUNICATIVE PHENOTYPES

A study by Didden (2009) identified and documented the communicative forms and functions in which children with Angelman Syndrome use naturally. Seventy-nine surveys were conducted and compared existing communicative skills in relation to the child's setting, genetic subtype, presence of epilepsy, age, environment, and level of intellectual disability. The results of this study suggest that the communicative phenotypes associated with Angelman Syndrome (e.g., disomy, epilepsy versus no epilepsy, the presence of epilepsy medication, severe versus profound level of intellectual disability, environmental factors, etc.) may be relevant to consider for any attempt to enhance the child's communicative functioning. This is significant because it provides insight that the communicative phenotypes associated with Angelman Syndrome influence the communicative forms and functions that exist naturally. This claim is corroborated by other articles. In the Summers and Impey (2011) reading, the children's chromosomal variation, the presence of seizures and whether the child is taking seizure medication, and their home environment are all taken into consideration before participants were selected, as well as included in the results. In the Didden et. al. (2009) article, the participants' age and chromosomal variation. Therefore, those genetic, medical, and environmental factors need to be considered in all intervention planning for children with Angelman Syndrome. This suggests that interventions designed specifically for people with Angelman Syndrome might benefit from alternative versions for the different varied communicative phenotypes.

BEHAVIORAL CHARACTERISTICS

Hyperactive behavior and impulsivity often cause difficulty with joint attention and communicative sharing, (Dan, 2008). Summers and Impey (2011) assessed the impairments in joint attention that are found among children with Angelman Syndrome. It included four children with Angelman Syndrome in ages five through ten years old. The children's joint attention responding was assessed using pictures and toys. The child's name was called to initiate eye contact and the examiner tracked eye movements of the child to establish attention span. If the child looked at the picture book within five seconds, the child was awarded a '3' and if eye contact had to be re-established by the examiner, the child would receive a '2'. If the examiner had to say "look" to get the child's attention, the child would receive a '1'. If the child did not look at the picture at all, the child would receive a '0'. A maximum score of 18 was not achieved by any of the participants, who respectively scored a 7, 15, 7 and 0. Gaze shifts occurred for all four children, and eye contact had to be re-established more often in 2 of the children. All four children were relatively less impaired in responding to the examiner's joint attention bids than in initiating joint attention with the examiner. The findings suggest that the children exhibited an early or more basic form of joint attention. This finding aligns with reports from other studies indicating that the acquisition of communication skills usually gets easier as children with Angelman Syndrome get older and their attention span increases, as attention span is a common limitation for children with Angelman Syndrome, (Clayton-Smith 2003). The results of this study concluded that understanding the nature of joint attention deficits in children with Angelman Syndrome may assist with developing approaches to improve their communication and social skills, (Summers & Impey, 2011).

ASSESSMENT

Didden et. al. (2009) compared the communicative functioning of individuals with Angelman Syndrome (n=109) with individuals with mixed etiologies (n=117) using the Verbal Behavior Assessment Scale (VerBAS). The results suggested that individuals with Angelman Syndrome had significantly lower scores on tacting and echoing, but not on manding. Manding refers to communication behaviors that produces a specific reinforcer and tacting produces a generalized reinforcer. Echoing is when an individual imitates other people's communicative behaviors, (Didden et. al., 2009). This indicates that people with Angelman Syndrome prefer specific reinforcers (manding) over generalized reinforcers (tacting). This suggests that when conducting intervention methods, Applied Behavior Analysis (ABA) techniques in particular, specific rewards garner better results and participation than general rewards do for children with Angelman Syndrome. This is important to note when proposing or conducting any study including individuals with Angelman Syndrome and wish to utilize rewards in a way that contributes positively to the study.

The VerBAS questionnaire is given to parents of people with developmental and intellectual disabilities in order to gain an understanding of their communication skills at home as well as at school. The parents and teacher will answer the questions to the best of their ability by rating on a scale from zero to five the ability as they see and understand the child's communication skills. The list of questions gives researchers a baseline of communication methods that are already achieved and can also help to determine whether a participant qualifies for their given study, depending on the desired answers to the listed questions, (Duker, 1999).

ASSISTIVE TECHNOLOGY

Children with Angelman Syndrome have been able to use augmented communication devices with success, and their acquisition of communication skills usually gets easier as they get older and their attention span increases, as attention span is a common limitation for children with Angelman Syndrome, (Clayton-Smith 2003). Calculator (2014), administered a survey to parents of children with Angelman Syndrome that gathered information about the children's current methods of communication, which included augmentative and alternative communication systems. Further exploration of children who used these communication systems measured the level of importance, usefulness, success, acceptance, and functionality with a rating scale as well as closed questions. In this survey, parents reported "Strongly Agree" and "Agree" far more often than "Strongly Disagree" and "Disagree" to questions such as: "My child uses the device efficiently" as well as, "This device has contributed positively to my child's overall quality of life," (Table 2). The results of this study suggest that there is clear evidence that augmentative and alternative communication device usage is often associated with positive outcomes, (Calculator, 2014). In the Didden et. al. (2009) reading, the authors found that about 30% of the individuals with Angelman Syndrome in their samples utilize some form on symbolic, non-speech, form of communication including picture exchange systems. These communication forms indicate that the use of augmentative and alternative communication devices is both an achievable goal and contributes positively to the children's overall communication skills.

IMPLICATIONS

When conducting a proposed research study on possible intervention methods for children with Angelman Syndrome, there are many aspects of the disease that need to be considered. The review of literature suggests that the specified type of chromosomal variation

(e.g., deletion, mutation, abnormalities, etc.), communicative phenotypes, attention span and joint attention, properly utilizing the preference for manding (specific reinforcers), and the preference for augmentative and alternative communication devices are all aspects that must be considered when conducting a thorough research study on acquiring communication skills in children with Angelman Syndrome. A study of this type was not located through the review of literature. Therefore, given this valuable information, the proposed research method will be conducted on how best to acquire functional communication skills in children with Angelman Syndrome using their preferred augmentative and alternative communication devices. This research will also include alternative versions of the principal intervention method that can be applied to any child with Angelman Syndrome within the previously noted considerations of the disease.

METHOD

EXPERIMENTAL DESIGN

The experimental design for this study will be a single-case multiple baseline design. This is a widely used approach for evaluating the effects of interventions on individuals. Multiple baseline designs involve repeated measurement of outcomes over time and the controlled introduction of a treatment at different times for different individuals. This approach to intervention research is common among general education and special education fields.

SETTING AND INTERVENTIONIST

The setting for this study will be the child's home environment, whether that is at home with parents/guardians or in a group home setting. The interventionist will be trained on what behaviors to expect, how to work one-on-one with the child, and how to accurately assess the

child's progress. See "Interventionist Training" for the procedures that the interventionists will practice.

PARTICIPANT

The proposed participants for this study will include no more than three children with a diagnosis of Angelman Syndrome at around ten years of age. Having three children in this study will allow for proper management as well as a varied, yet concise, result. The children's chromosomal testing and diagnostic criteria will be reviewed in order to establish what chromosomal variation of Angelman Syndrome the child has, as well as any communicative phenotypes (epilepsy versus no epilepsy, severe versus profound level of intellectual disability, environmental factors, age, etc.) that are present. These differences will be noted within the context of the study in order to ensure that the proper intervention techniques are being utilized. The age of around ten years old shall include no child under nine and no child over eleven years old. This age range is chosen because younger children with Angelman Syndrome's extremely short joint attention span makes one-on-one intervention methods less successful, (Summers & Impey, 2011). At about ten years of age their attention spans begin to lengthen, allowing for more interaction and more successful interventions, especially for acquisition of communication skills**.**

TECHNOLOGY AND MATERIALS

Materials required for this study include: three augmentative and alternative communication devices, a list of communicative phenotypes associated with the specific child, a scoring sheet for the interventionist to fill out for both establishing a baseline and the execution of the intervention method.

MEASURES

VerBAS

The data on communicative functioning will be collected using VerBAS. Values range from '0' (never) to '4' (always) and are based on frequency of occurrence based on parental/guardian and teacher assessment. Maximum score of each subscale is 20 and the maximum total score of the VerBAS is 60, indicating the overall level of expressive communicative functioning. This is done to establish that the child uses communication at a basic level and is a good candidate for the intervention. After the assessment is completed and analyzed, the intervention will begin.

TASK PROBE

The interventionist will count how many trials are done and how many successful attempts are achieved using the scoring sheet. This will allow for a consistent measure of success across each child that can be compared in order to properly analyze the results of the study. The interventionist will use the scoring sheet to indicate whether the child's ability to use the communication device after the intervention has been completed is 'present', 'partially present', or 'not present'. There will also be a 'notes' section for the interventionist to include any information that they believe is pertinent to the study. See Figure 1 for the Scoring Sheet.

PROCEDURE

INTERVENTIONIST TRAINING

Interventionists in this study will be trained on ABA best practices and be qualified to work with children with severe special needs. The interventionist will be taught not to use "filler words" such as "the" and "and" when conducting the intervention and establishing a baseline.

This is because, for the purpose of intervention, the child should focus on the instructional words without having to decipher and decode more words than is necessary.

BASELINE CONDITION

When establishing the baseline for every participant, the interventionist will use the augmentative device to administer the sessions before the intervention method begins. This will give the interventionist a starting point to measure where the child's communication skills were both at the start and the end of the intervention. This helps the researcher gain an understanding of what the impact of the intervention is.

When conducting the intervention method, the interventionist will introduce the child to the device, and use a physical object to relate it to the device. For example, the interventionist will say "Want cup?" and will press the button with the picture of a cup. When the device says the word "cup" the physical cup will then be handed to the child. When the child hands back the cup, the interventionist will ask again "Want cup?" and give a few attempts for the child to press the cup button, indicating that yes, they want the cup. After the child presses the button successfully, it is important that the interventionist praise the child, clap, and hand them the cup. Because children with Angelman Syndrome are very sociable and thrive with attention, this is a great reinforcer to utilize, (Dan, 2008). Ten trials of this will be repeated until ninety percent success is achieved. Then another photo, this time of a toy that the child enjoys, will be used to replace the cup, indicating to the child that this method of communication can be generalized. Throughout the length of this intervention, the child should be able to use at least three icons on the device proficiently (ninety percent success).

INTERVENTION DESCRIPTION

The script that will be provided to the interventionist will include the following:

Step One:

Start the recorder, say the date and trial number.

Introduce the photo on the device to the student and relate it to the object inside the home.

Say, "*Cup*" and press the photo of the cup on the device. Wait for the device to repeat the word, then hand the cup to the child.

Say, "*Cup*" again and use the student's hand to press the photo of the cup. Wait for the device to repeat the word, then hand the cup to the child.

Ask "*Want cup*?" and point to the picture on the device.

Give three attempts for success.

When the device is used successfully, show excitement and praise, and hand the student the cup.

Do ten trials.

Record the amount of successful trials versus unsuccessful trials on the Scoring Sheet.

Step Two:

After two days of 100% success, a new photo and object should be introduced to be used along with the previous one.

Start the recorder, say the date and trial number.

Introduce the **new** photo on the device to the student and relate it to the **new** object inside the home.

Say, "*Toy*" and press the photo of the toy on the device. Wait for the device to repeat the word, then hand the cup to the child.

Say, "*Toy*" again and use the student's hand to press the photo of the cup. Wait for the device to repeat the word, then hand the toy to the child.

Ask "*Want toy*?" and point to the picture on the device.

Give three attempts for success.

When the device is used successfully, show excitement and praise, and hand the student the cup.

Do ten trials.

Record the amount of successful trials versus unsuccessful trials on the Scoring Sheet.

Step Three:

Continue to introduce new objects with their corresponding photos, up to five photos. Make sure the photos are accessible to the student as needed, and that the correct photo is being used for the correct object. When the child correctly indicates a request or statement on the device for five pictures with ninety percent accuracy, the child has achieved success with their device.

FIDELITY OF IMPLEMENTATION

The data that will be used to ensure the interventionist adhered to the steps of the intervention is an audio/video recording of the baseline session as well as the intervention session. The interventionist will also be given a form that outlines the instructions and script for them to follow, as well as a scoring sheet that will be reviewed after every session.

DATA ANALYSIS

According to Kratochwill, et. al. (2010), single-case design studies have many variations, and these designs often involve, "repeated, systematic measurement of a dependent variable before, during, and after the active manipulation of an independent variable (e.g., applying an intervention)," (p. 2). The visual analysis in single-case design studies consist of four steps and six variables. The first step is documenting a baseline pattern that is predictable. Once that is established, the second step is to assess if there are sufficient data with enough of a pattern to establish a pattern. The third step is to compare the data from each phase with the data in an adjacent phase to assess whether manipulation of the independent variable was associated

with a predicted change in the pattern of the dependent variable. The fourth step is to integrate all the phases of the study to determine whether there are at least three significant changes at different points in time, (Kratochwill et. al., 2010). The six variables involved are, "level, trend, variability, immediacy of the effect, overlap, and consistency of data patterns across similar phases," (Kratochwill et. al., 2010, p. 18).

STRENGTHS AND LIMITATIONS

Limitations of this study are scope and time. This is due to the limited amount of time and the small number of participants. A longitudinal study that includes various children of varying ages would be the best implementation of this study, but this is not ideal at this time due to a limitation of time and resources.

A strength of this study is the process in which a baseline will be established in order to ensure the participants are improving their communication skills in line with the expectations of the study. Another strength is the inclusion of the communicative phenotypes in relation to communication, as not many studies have included this in their methodology.

CONCLUSION

This proposed study will be conducted on how best to acquire functional communication skills in children with Angelman Syndrome using their preferred augmentative and alternative communication devices. The literature suggests that the specified type of chromosomal variation (e.g., deletion, mutation, abnormalities, etc.), communicative phenotypes, attention span and joint attention, properly utilizing the preference for manding (specific reinforcers), and the preference for augmentative and alternative communication devices are all aspects that must be considered when conducting a thorough research study on acquiring communication skills in children with Angelman Syndrome.

Given this information, this study will be using a single-case multiple baseline design. Three children with Angelman Syndrome will receive high-quality intervention that can be used to inform future research on acquiring functional communication skills using augmentative and alternative communication devices for children with Angelman Syndrome. The implications of this study suggest that functional communication skills are achievable for children with Angelman Syndrome.

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Figure

Figure 1:

Criteria	Present	Partially Present	Not Present	Notes
Was the child attentive to the device?				
Did the child press icons successfully?				
Did the child initiate use of the device?				
Did the child press the icons independently?				
Did the child press icons with assistance?				
# of icons the child able to use proficiently?				#
Can the child use the device with 90% accuracy?				
How many attempts did it take to achieve success?				