

Highlighting Speech and Language Characteristics in Angelman Syndrome: A Case Study

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Abstract:

Introduction

Angelman syndrome is a rare genetic neuro-developmental disorder diagnosed in one in 12000-20000 live births (NORD, 2018). Individuals with Angelman syndrome typically present with global developmental delay, learning difficulties, intellectual disability, seizures, ataxia, and a distinctive behavioral phenotype (Williams, 2010). One of the hallmark features of Angelman syndrome is severe speech and language impairment.

Aim

The study aimed to highlight speech and language characteristics in a case with Angelman syndrome.

Methodology

A case aged four- year Hindi speaking female child who reported with chief complaint of unable to speak clearly. Comprehensive Speech and language evaluation included administration of Receptive-Expressive Emergent Language Scales (REELS) (Kenneth R Bzoch), Communication Developmental Eclectic Approach to Language Learning (Com-DEALL developmental profile) (Pratibha Karanth, The Com-DEALL Trust), Speech intelligibility rating scale (AYJNIHH-7 point rating scale), Photo Articulation Test -Hindi (UNICEF project) , Com-DEALL Oro-motor Checklist (Pratibha Karanth, The Com-DEALL Trust) from a SLP perspective. The test scores were collected from the child's behavior observation during clinical set-up and parental responses from daily life.

Result and discussion:

The preset study result indicated delayed developmental scores in receptive and expressive language, fine motor skills and cognition. Speech errors are noted in the domain of articulation. Post therapeutics findings indicated significant change in the developmental scores of all the domain.

Conclusion:

Research on assessment and intervention protocol of Angelman syndrome can be a stepping stone in terms of exploring information to provide adequate awareness and counseling of parents.

Keywords: Angelman syndrome, Com-DEALL, Global developmental delay

Introduction:

In 1965 Harry Angelman, a British pediatrician, described the "Puppet Children," later being renamed Angelman syndrome. It is a classic example of genomic imprinting, where the expression of a genomic region differs depending on the chromosome's parent of origin. The Angelman syndrome (AS) is a genetic disease that is considered uncommon. This syndrome is caused by the lack of expression of the chromosomal region's imprinted genes. In the case of AS, the loss of information takes place in the maternal chromosome. (Ehrhart et. al, 2018). The estimated prevalence in children and young adults between 1 in 10,000 and 1 in 20,000 (Williams, 2005). The incidence of Angelman syndrome (AS) varies from 1 in 20,000 to 1 in 12,000 live births. There is no gender preference, and AS affects males and females equally. However, many cases may go undiagnosed due to the underreporting of cases and misdiagnosis (Fiumara, 2010).

Angelman syndrome (AS) is characterized by severe developmental delay or intellectual disability, short attention span, severe speech impairment, gait ataxia, tremulousness of the limbs, and unique behavior with an apparent happy demeanor that includes frequent laughing, smiling, and excitability. Microcephaly and seizures are also common. Developmental delays are first noted at around age of six months; however, the unique clinical features of AS do not manifest until after age one year. Speech development issues, ranges from being non-verbal to only using a few words for communication. Common facial features of Angelman syndrome include macroglossia, mandibular prognathia, widemouth and widely spaced teeth (Dagli et. al, 2021).

Angelman syndrome is a rare neuro-developmental disorder with a genetic basis in maternal genomic imprinting that denotes behavioral and cognitive impairment. The presentations of developmental delay in Angelman syndrome may be vague in early life, resulting in the potential delay in diagnosis and intervention. Angelman syndrome represent with initial diagnostic confusion with multiple associated pathologies. Hence the present case study aimed at a holistic speech and language evaluation. The main complications arising in Angelman syndrome (AS) individuals are due to their comorbidities such as seizure and ataxia; the patient can have injuries during these episodes. However, AS leads to an increased risk of accidents due to the combination of hyperactivity, exploratory behavior, and intellectual disability.

Methods

A case aged four- year Hindi speaking female child who reported at Speech and Hearing department with chief complaint of unable to speak clearly. Case history revealed that the child had a history of seizure at the age of 2 years. *The clinical impression on pediatric examination indicated the presence of characteristics of Angelman syndrome.* The medical report depicted occasional right fronto-central epileptiform discharge. Delayed motor developmental milestone was reported based on parental perception. The child communicated pre- dominantly through mainly phrases and occasionally in simple sentences. Macroglossia and wide mouth facial clinical features noted. Behavior observation and parental perception indicated an apparent happy demeanor that included frequent laughing, smiling, and excitability.

Procedure: Comprehensive Speech and language evaluation included administration of Receptive-Expressive Emergent Language Scales (REELS) (*Kenneth R Bzoch*), Communication Developmental Eclectic Approach to Language Learning (Com-DEALL developmental profile) (Pratibha Karanth, The

Com-DEALL Trust), Speech intelligibility rating scale (AYJNIHH-7 point rating scale), Photo Articulation Test-Hindi (UNICEF project) , Com-DEALL Oro-motor Checklist (Pratibha Karanth, The Com-DEALL Trust) from a SLP perspective. The test scores were collected from the child’s behavior observation during clinical set-up and parental responses from daily life.

TREATMENT Procedure: The speech and language intervention guidelines in the present case of *Angelman syndrome* highlighted on hybrid approaches. Frequency of the therapy sessions were set thrice in a week for 45 minutes per session. Treatment goals were focused on (1) To enhance language using modeling, extension and expansion techniques. (2) To work on correction of articulatory errors. (3) To enhance cognitive learning through clinical and home settings observation. (4) To facilitate strengthening exercises for oral peripheral structures.

To facilitate language list of familiar and unfamiliar items was used for identification and naming of different lexical categories. Verbalexpression through sentences was also stimulated in structured and unstructured situations. Cognition was improved by using interactive vocabulary book for the recalling counting and multistep directional commands.

To facilitate oral peripheral structures different size of straws, whistles, thermocol balls, candle blow and thermal stimulations were used.

Result:

The current study aimed to highlight on assessment and intervention helps to improve the child’s different skills for communication in daily life situation. A detailed speech and language evaluation was done using standardized tests in structured and unstructured sessions. Receptive-Expressive Emergent Language Scales (REELS) revealed the receptive language age and expressive language as 3-3.5 years and 27-30 months respectively. *COMDEALL checklist* was administered to assess different developmental domains such as (gross motor (GM) Fine motor (FM), Activity of daily living (ADL), Receptive language (RL), Expressive language (EL), Cognition (Cog), Social (Soc), Emotional (Em).

Table: 1 Age Range in COMDEALL Checklist

Domain	GM	FM	ADL	RL	EL	Cog	Soc	Em
Age	48-42	42-36	48-42	48-42	24-30	36-30	42-36	48-42

Graph 1 Profile of age range in COMDEALL Checklist

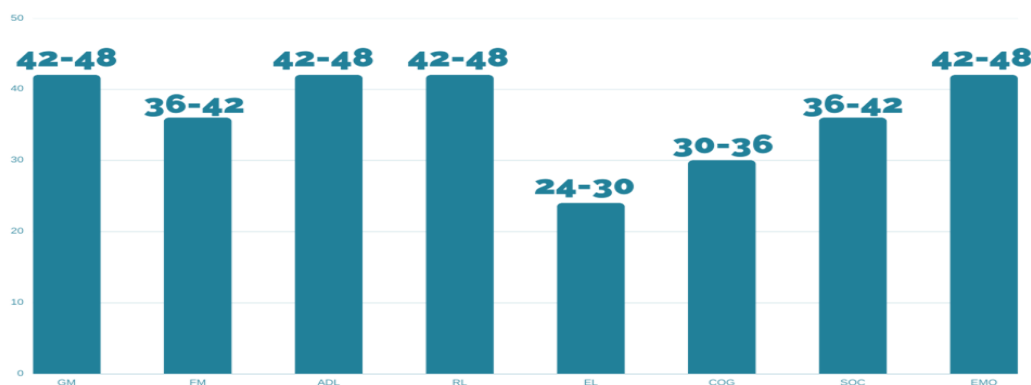


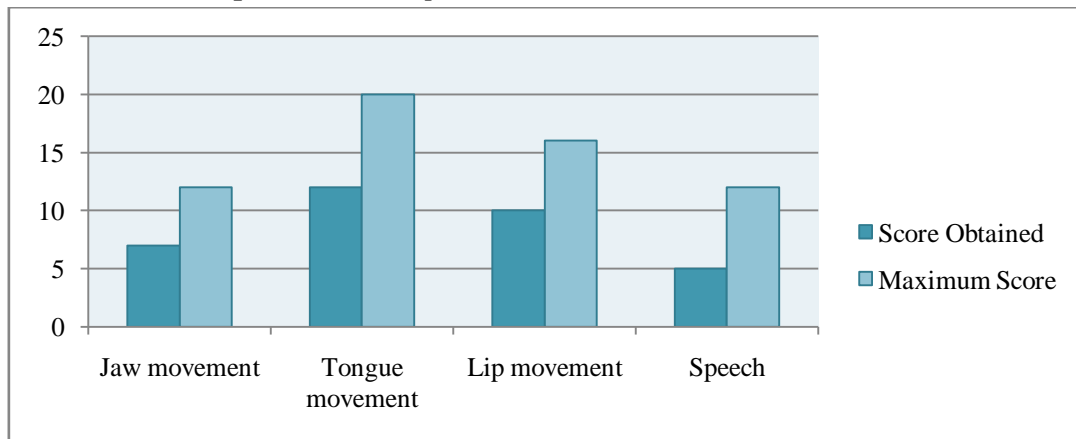
Photo Articulation Test (PAT-H) in Hindi was administered and results revealed substitution errors of velar, retroflex, fricative and affricates at all position. Pre-dominant omission of blends was present at

word level. Speech Intelligibility Rating Scale (AYJNIHH-7 Point) was done using picture card description and conversation during intervention session suggested that child’s speech is difficult to understand with many words unintelligible.

Table: 2 Oral Peripheral Structures Examination scores (ComDEALLOro Motor Checklist)

Section	Score Obtained	Maximum Score
Jaw movement	07	12
Tongue movement	12	20
Lip movement	10	16
Speech	05	12

Graph 2 Oral Peripheral Structures Examination scores



The above table and graph shows the score of COMDEALL assessment of oro motor skills in toddlers which imply oro motor skills deficiency.

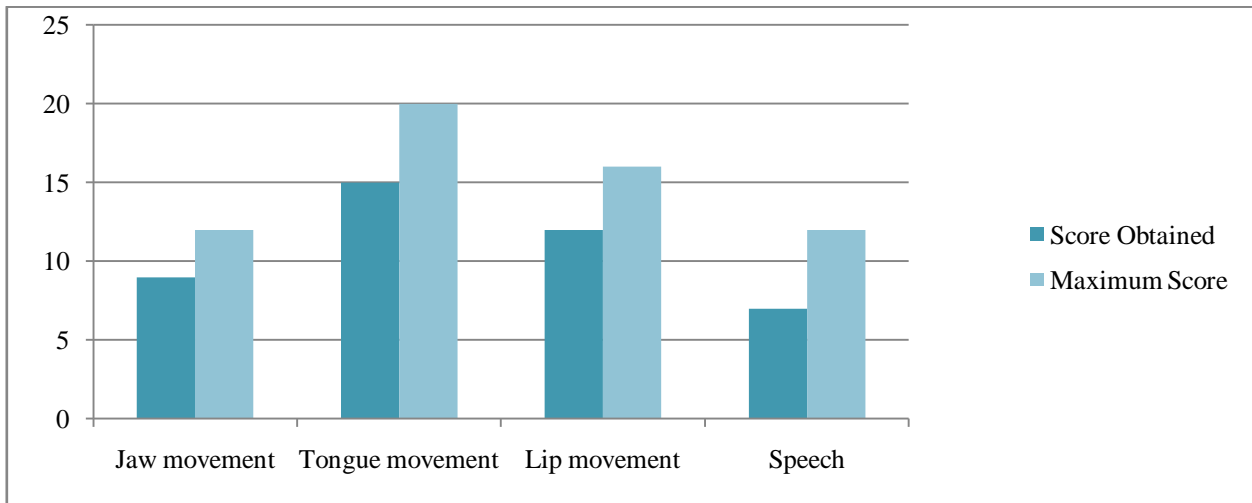
Detailed speech and language evaluation and after intervention of 24 sessions, performance on speech, language, cognition and oral peripheral structural exercises suggested improvement with spontaneous response in language, cognition task while inconsistent response seen in expressive speech task. To obtain discrete findings for all affected domains, therapy sessions were monitored to see the improvements. After 24 sessions re-evaluation was done to assess the progress.

Post intervention scores of Receptive-Expressive Emergent Language Scales (REELS) revealed the receptive language age and expressive language as 3.5- 4 years and 33-36 months respectively.

Table: 3 Post-Therapeutics Oral Peripheral Structures Examination scores

Section	Score Obtained	Maximum Score
Jaw movement	09	12
Tongue movement	15	20
Lip movement	12	16
Speech	07	12

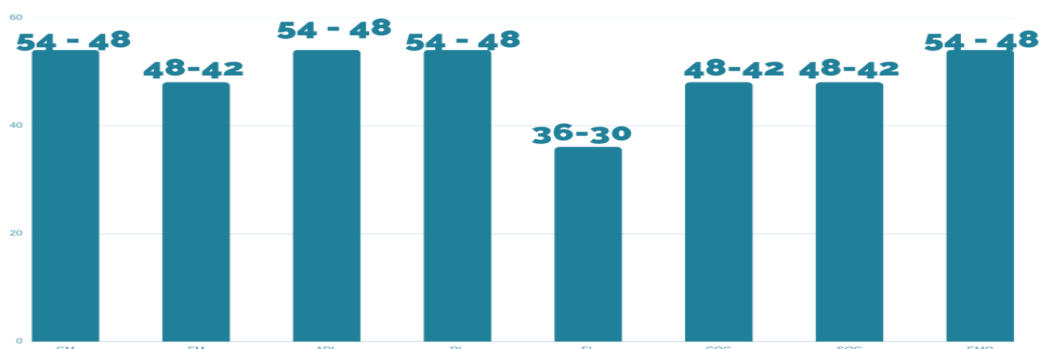
Graph 3 Post-Therapeutics Oral Peripheral Structures Examination scores



The above table and graph indicates that verbal and non-verbal tasks related to oral peripheral structures shows significant improvement and increased strength in all active articulators.

Table: 4 Post interventions Age Range in COMDEALL Checklist

Domain	GM	FM	ADL	RL	EL	Cog	Soc	Em
Age	54 - 48	48-42	54 - 48	54 - 48	36-30	48-42	48-42	54 - 48



Graph 4 Profile of age range in COMDEALL Checklist

The results of the post therapeutic ComDEALL is in support of the study of (Grieco, 2018) which stated that Communication is extremely restricted compromised and are slowly acquired as pre-intentional behaviors do not progress typically. Communicative and linguistic deficits are attributed to intellectual changes, which hinder the acquisition of communicative abilities, and motor changes.

Photo Articulation Test (PAT-H) in Hindi assessed post therapeutically after 24 sessions revealed improvement in articulation of velar, & fricative sounds in isolation, and at initial word level. Slight improvement was noticed in overall speech Intelligibility

The articulatory errors registered in this case study are supported by (Dagli et. al, 2021) which stated that AS children have severe speech impairment.

Conclusion:

Assessment of speech and language skills in Angelman Syndrome showed delayed in speech, language, cognition and motor skills. Formal therapy plan helped in enhancement of the affected domains in this present case study. Research on assessment and intervention protocol of Angelman syndrome can be a stepping stone in terms of exploring information which in-turn helps in providing adequate awareness and counseling of parents.

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