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MOVEMENT MILESTONES – "HOW PHYSIOTHERAPY ENHANCED MOTOR ABILITIES IN ANGELMAN SYNDROME – A CASE REPORT"

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ABSTRACT

Background: Angelman syndrome (AS) is a rare, neurogenic disorder signs of global developmental delay, profound intellectual disability, ataxia, and epilepsy (found in approximately 80% of cases). Other notable characteristics includes absence of speech, distinct behavioral patterns, sleep disturbances, a cheerful disposition with frequent laughter, and hyperactivity. Evidence-based physiotherapy protocols are increasingly used to manage deficiencies in trunk control, mobility, balance, and gait in individuals with AS.

Aim: To evaluate the effect of a targeted physiotherapy protocol on motor function in a child with Angelman syndrome.

Case Description: This case report details a 7-year-old girl with AS with significant challenges in gross motor activities like independent trunk control, sitting and standing.

Methodology: A comprehensive & targeted physiotherapy regimen was designed using ICF model. Evidence based intervention trunk stability, mobility and balance was administered over 36 sessions. The treatment regimen involved trunk stability & mobility exercises, progressive balance exercises, standing and gait training with support. The Gross Motor Function Measure (GMFM-88) focusing the C, D, and E Dimensions was used to evaluate the functional changes.

Conclusion: Findings from this case study indicate that a structured and targeted exercised can improve the functional motor performance inspite of various system level constraints in children with Angelman syndrome.

Keywords: Happy puppet, mobility, trunk control, Pediatric neurogenic disorders

INTRODUCTION

Angelman Syndrome (AS) is a rare, incurable neurodevelopmental disorder caused by the loss of function in the maternally inherited UBE3A gene, located within the 15q11-13 region of chromosome 15 **[1, 2]**. This gene encodes the ubiquitin-protein ligase E3A, essential for proper neuronal function through the ubiquitin-proteasome pathway, which regulates protein degradation in the brain. The absence of functional UBE3A gene activity occurs due to gene deletion, mutation, imprinting defects, or paternal uniparental disomy (PUD) in neuronal function leads to a range of symptoms that define AS **[3]**. AS affects approximately 1 in 20,000 individuals, with both males and females equally affected **[4]**. The most common genetic cause, accounting for 70-75% of cases, is a deletion in the maternal UBE3A gene on chromosome 15q11-13. Other causes include PUD (2-3%) and imprinting center defects (3-5%). Symptoms typically observed around six months of age and primarily affect the nervous system, leading to severe global developmental delay, intellectual disability, ataxia, epilepsy (in 80% of cases), absence of speech, distinct behavioral traits, sleep disturbances, frequent laughter, and hyperactivity **[5]**. Diagnosis of AS involve genetic testing, often through DNA sequencing, to identify mutations in the UBE3A gene, as well as EEG to detect characteristic epileptic patterns. Although no cure currently exists, symptomatic management can improve quality of life and functional abilities in children with AS **[4]**. This case study aims to evaluate the effectiveness of a targeted physiotherapy program designed to improve motor function, particularly focusing on core stability, balance, and gait in AS.

CASE PRESENTATION

A comprehensive assessment was conducted by obtaining relevant history and symptoms as reported by the child's grandmother, who brought her for the therapy sessions. The parents had a non-consanguineous marriage, and no other family members are reported to have Angelman Syndrome. The mother attended all routine prenatal examinations consistently and adhered to her prescribed medications. Due to oligohydramnios, she underwent an emergency lower segment cesarean section (LSCS) and delivered one week before the anticipated due date. The child cried immediately post-birth and remained in stable health until six months of

age. However, at seven months, she experienced a high fever followed by seizures. By nine months, after consultations with several physicians, the child was referred to super specialty Hospital, where a genetic test for copy number variation (CNV) revealed a deletion of the UBE3A gene, confirming a clinical diagnosis of Angelman Syndrome.

CLINICAL FINDINGS

The assessment was based on her dialogue, detailing that the child could roll from supine to prone and transition independently into a quadruped position but was unable to initiate standing from a seated position. The child demonstrated head control by four months of age and achieved rolling from supine to side-lying by the fifth month. By the sixth month, she was able to roll from supine to prone and from prone to supine. Sitting with support was achieved after two years of age. Attempts to stand with support were first observed after three years and by four years, the child could take few steps with support. However, independent walking and stair climbing have not yet been achieved. Fine Motor Development as voluntary grasp and reaching were observed by the fifth month, while pincer grasp was developed after one year. The child demonstrated a social smile by the second month. Monosyllabic and bisyllabic verbalizations have not yet been achieved. The child is currently on anti-epileptic medication and takes sleeping medication to manage nocturnal disturbances. Sleep patterns indicate that the child sleeps only four hours per night despite medication, with drowsiness observed during the day. She exhibits incontinence of both bowel and bladder and follows a mixed diet.

During sessions, the child displayed spontaneous laughter without obvious cause and was often drowsy, irritable, and occasionally uncooperative. She had an ectomorphic body build and can maintain a sitting posture with a mild kyphotic curvature. No abnormal limb positioning was noted; however, in a supine position, both lower limbs displayed excessive external rotation. Observed involuntary movements included intermittent shivering of the upper limbs and hand flapping. The child's was dependent and was carried by the caregiver. Physical examination showed a head circumference of 45 cm and passive full range of motion (ROM) in all degrees of freedom for both upper and lower limbs, without any deformities. Higher Mental Function examination showed the child was conscious but not fully oriented. She recognizes family members, responds to play activities, but her communication abilities are impaired, as she is unable to speak. Superficial reflexes, including abdominal, corneal, and plantar reflexes, were present. Deep reflexes, such as the patellar and biceps reflexes, were also

present. Primitive, spinal, and brainstem reflexes were integrated.

The child has normal muscle tone and no spasticity. Muscle power assessment using manual muscle testing (MMT) could not be completed due to her lack of orientation. Muscle girth measurement dint show any significant variations. On observation the child was achieving progressively trunk stability and hence static balance was minimal. Further the child was completely dependent for ambulation. The child's motor development was evaluated using the Gross Motor Function Measure (GMFM-88), assessing components relevant to the child's abilities. The child had achieved Component A (lying and rolling) and demonstrated independent sitting for Component B. For Component C (crawling and kneeling), the child could crawl only short distances and was unable to reach a kneeling position. Component D (standing) was achieved only with support, and for Component E (walking, running, and jumping), the child could initiate walking but required assistance. International classification for functioning, disability and health (ICF) model was used to understand the level of capacity & performance & towards goal setting **(Table 1)**.

Table 1: Shows ICF model for goal setting

Body structure and functional limitation	Activity limitation	Participation restriction
• Core muscle weakness • Lack of trunk stability • Lack of trunk mobility • Lack of balance. • Lack of postural control. • Impairment in gait.	• Standing • Walking • Unable to go to the washrooms • Reduced gross motor activities.	• Reduced family participation. • Unable to attend school Constrained higher mental function • Impaired school education & participation in peer group • Lack of socialization
Environmental and personal factors		
Facilitators • Family support • Access to therapy	**Barriers** • Drowsy • Lack of involvement & motivation • Anxiety	

Based on the above assessment, a targeted physiotherapy treatment plan was developed to address the identified issues, including trunk instability, poor balance, and inability to stand and walk independently using ICF model. Short term goals were to improve the trunk stability and strengthen the core muscles. And long term goals were to achieve stability and balance in standing, thereby to attain assisted walking.

INTERVENTION

Following a noticeable delay in developmental milestones, the child underwent physiotherapy at an out-patient care at the age of two, where interventions enabled her to achieve independent sitting. At seven years, she was brought to physiotherapy OPD at tertiary care hospital by her grandmother for further intense care. Written informed consent was obtained from the mother, and the treatment procedures were thoroughly explained prior to

commencing the protocol. After reviewing the literature **[6-10]** on AS, evidence based exercises were ensured. The protocol was delivered as three sessions per week on alternate days over period of three months, totaling 36 sessions. Each session lasted for one & half hour, with five-minute rest intervals between exercises. Progressive adjustments were made to exercises over time, with an increase in sets from two to three by the 20th session, as per the child's cooperation and mood manners. Visual stimuli & consistent dialogue with the child were employed to promote cooperation, and involvement. The exercises performed in this case study were delivered progressively as the child achieved the phase wise milestones **(Table 2)**. Each exercises were repeated for 15-20 repetitions.

Table 2: Shows the targeted exercise protocol delivered progressively

Exercise aim & goal	Types of targeted exercises		
Phase 1 **Foundation & Stabilization** **Primary goal:** **To build core stability and postural control, improve upper limb coordination, and introduce sensory integration**	**Core Strength and Trunk Control**	**Sensory Integration**	**Upper Limb Strengthening**
	A supported sitting device / bolster used to facilitate trunk control such as reaching to the sides or twisting. **The child was placed in prone /seated on a large therapy ball with support, promoting balance reactions by gently rolling or tilting the ball.**	**Tactile Stimulation was done using textured materials (e.g., soft fabric, sensory balls) to stimulate the child's hands and feet, enhancing proprioception.** **Vestibular Activities were incorporated by gentle rocking or swinging motions to stimulate the vestibular system, promoting spatial orientation and balance.**	**Supported reaching facilitated by positioning the child to reach for colorful, light-weight objects, encouraging shoulder and trunk activation.** **Resistive Grasping was introduced by hand-strengthening toys (e.g., squeeze balls, putty) for fine motor improvement.**
Phase 2 **Progressive Strengthening and Mobility** **Primary Goals:** **To increase the complexity of core and limb exercises, focus on weight-bearing activities, continue sensory-motor integration.**	**Core Stability and Sitting Tolerance**	**Lower Limb Strengthening and Coordination**	**Upper Limb and Visual-Motor Integration**
	Supported Weight Shifts were performed in a seated or kneeling position. The child was encouraged to perform weight-shifting exercises by reaching for toys placed at various heights. **Tilt Board Exercises with assistance performed by placing the child on a tilt board while seated, stimulating postural responses as they adjust to the board's movements.**	**Supported standing with a standing frame was introduced to promote controlled weight-bearing and gradually increasing as tolerated** **In a supine position, assisted leg lifts, bridging, and supported cycling movements were given.**	**Interactive Reaching Games with interactive tools, a simple touch screen device / switch-activated toys used to encourage reaching and controlled grasping, fostering hand-eye coordination.**
Phase 3 **Functional Integration and Dynamic Movement** **Primary Goals:** **To Maximize functional movement patterns, promote balance reactions, refine sensory integration, and encourage independent upper and lower limb coordination.**	**Dynamic Core and Postural Control**	**Strength and Weight-Bearing Activities**	**Interactive Play with Sensory Equipment**
	Different seating positions (e.g., floor mat, therapy bench) used to challenge the child's ability to maintain sitting balance while reaching. **Core strength facilitated on therapy Ball and the difficulty level increased by encouraging gentle bouncing or reaching while on the ball.**	**Supported standing with standing frame, encouraged for reaching toys placed at various angles, stimulating balance and weight shifts.**	**Obstacle course play using low barriers like cushioned objects encouraged crawling, sliding movements, building strength and coordination.** **Vestibular swings and textured pathways were used for further sensory input, to engage visually and physically in novel ways.**
	Assisted walking movements	**Adaptive gait training**	**Sensory Integration**

Phase 4 **Progression to supported walking and gait pattern initiation** **Primary Goals:** **To introduce partial weight-bearing gait movements, and develop neuromuscular control in the legs.** **To enhance coordination for functional gait training.**	**Partial weight-bearing treadmill training with appropriate harness support, introduced at a slow pace, focusing on achieving rhythmic stepping.**	**Over ground assisted walking with manual assistance, focused on step placement and alternating limb movements.** **Lightweight ankle cuffs of ½ kg provided gentle resistance during assisted stepping on treadmill sessions, promoting strength in the hip flexors and extensors.**	**Incorporated textured mats to promote foot sensory stimulation and e weight shifting and stepping encouraged.**

The effect of the tailored and supervised exercises was examined using GMFS -88 and the observations are shown in **Table 3 & 4**.

Table 3: Shows the changes in dimension C, D, E of GMFS at before and after targeted physiotherapy

S. No.	Dimensions assessed	% of scores before intervention	After intervention
1.	Crawling & kneeling	50%	64%
2.	Standing	10.2%	20.5%
3.	Walking, running & jumping	8.3%	16.6%
4.	Total score	48.3%	55%
5.	Goal total score	23%	34%

Table 4: % of changes in motor function analyzed using GMFM-88

	GMFM -Dimension C (crawling & kneeling)			
S. No	Components	Pre-intervention	Post-intervention	Result
1.	Child is able to reach forward from crawling with right arm above the shoulder level	1	3	From initiates to complete.
2.	Child is able to reach forward from crawling with left arm above the shoulder level.	1	3	From initiates to complete
3.	Crawls forward 1.8m	2	3	Partially completes to complete.
4.	Able to crawls up using hands and knee	1	2	Initiates to partially completes
	Dimension D (standing)			
1.	Maintain in standing position arm 3 seconds for 3 seconds	1	1	No improvement
2.	Child holds to the large bench with one hand support and lifts right leg for 3 seconds.	0	2	Not initiate to partially completes
3.	Child holds to the large bench with one hand support and lifts left leg for 3 seconds.	0	2	Not initiate to partially completes
	Dimension E (walking, running and jumping)			
4.	Standing on large bench with both hand support and moves forward 5 steps to the right side	0	2	Not initiate to partial complete
5.	Standing on large bench with both hand support and moves forwards 5 steps to the left side	0	2	Not initiate to partial complete
6.	Standing with one hand support and walks forward 10 steps	2	3	Partially complete to fully complete
	Total scores of 3 domains	8	23	-

DISCUSSION

The case of a 7-year-old child with AS who showed clinically improved changes in gross motor function following a tailored physiotherapy program over three months provides valuable insights into the potential benefits of targeted rehabilitation interventions for this population. Angelman syndrome is characterized by severe motor and cognitive developmental delays, which

often result in reduced activity and participation levels.

One notable study reported on the progress of a 7-year-old boy with AS who underwent a synergic intervention involving translingual neurostimulation and goal-oriented rehabilitation. This intervention led to considerable improvements in spatiotemporal and kinematic gait variables, as well as a significant increase in walking distance from 500 meters to 2 kilometers over a four-week period. This case underscores the importance of innovative and intensive rehabilitation strategies in enhancing motor abilities in children with AS **[11]**. Similarly, another study investigated the effects of a three-year physiotherapy program on a child with AS, utilizing the Neurodevelopmental Treatment approach. The results demonstrated substantial improvements in gross motor function, with the GMFM scores increasing from 11.46% to 70.82% **[12]**. These findings highlight the effectiveness of sustained physiotherapy interventions in improving motor performance and overall functional abilities in children with AS2. This current case study explored how targeted interventions might promote functional gains. Developmental delays in AS typically emerge between 6 to 12 months of age; in this case, the child presented with early developmental issues, with multiple seizures, and sleep disturbances, prompting targeted therapeutic intervention. During the three-month intervention period, therapy focused on addressing the child's primary limitations, including trunk instability, reduced mobility, and functional activity restrictions. Evidence-based interventions included trunk stability and mobility exercises, static and dynamic balance activities, and treadmill training to support gait development.

Baseline GMFM-88 scores in standing & walking were low, indicating considerable motor impairment. Following focused and tailored PT intervention, changes in scores of standing and walking increased by 50%. These increases, though modest, reflect a positive trend in motor function, consistent with evidence-based findings that targeted physiotherapy can improve functional motor outcomes. The tailored physiotherapy program in the present case report likely contributed to the observed improvements in gross motor function, enabling the child to sit and walk independently. The positive outcomes observed in this case are consistent with the findings from the aforementioned studies, which collectively emphasize the potential benefits of targeted and intensive rehabilitation interventions for children with AS. These interventions not only enhance motor abilities but also improve overall participation and quality of life.

Challenges encountered during the study included drowsiness due to daily sleep medication, immune-related issues causing frequent absences, and occasional noncompliance due to agitation. Additionally, the limitations of available outcome measures posed challenges in fully quantifying motor improvements. However, despite these limitations, this case study underscores the potential value of physiotherapy in supporting motor function in AS, offering qualitative insights into the intervention's effectiveness.

CONCLUSION

The case of the 7-year-old child with AS demonstrates the significant impact that a tailored physiotherapy program can have on gross motor function. The findings from this case, along with supporting evidence from other studies, underscore the importance of developing and implementing targeted rehabilitation strategies to address the unique needs of children with AS. Further research is warranted to explore the long-term effects of such interventions and to identify the most effective approaches for optimizing motor function and participation in this population.

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Conflict of interest

The authors declare no potential conflict of interest

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