

# Impact of Functional Activity-Based Physiotherapy with NDT Approach on Motor Performance and Functional Independence in a Child with Angelman Syndrome: A Case Study

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## ABSTRACT

Angelman syndrome (AS) is a rare neurogenetic disorder characterized by severe developmental delay, motor incoordination, epilepsy, and distinctive behavioral features. Evidence supporting the effectiveness of physiotherapy interventions in AS remains limited. This case study examines the impact of a structured functional activity-based physiotherapy program incorporating Neurodevelopmental Treatment (NDT) principles on gross motor performance and functional independence in a 10-year-old female child with AS. Outcome measures included the Gross Motor Function Measure (GMFM) and Wee Functional Independence Measure (WeeFIM), assessed before and after a 4-week intervention. Post-intervention results demonstrated improvement in GMFM scores from 54.46% to 60.82% and enhanced WeeFIM scores, indicating gains in gross motor abilities and functional independence. The findings suggest that structured, goal-oriented physiotherapy can yield meaningful functional improvements even over a short duration in children with Angelman syndrome.

**Keywords:** *Angelman syndrome, physiotherapy, GMFM, WeeFIM, functional activity, NDT*

## INTRODUCTION

Angelman syndrome (AS) is a rare neurogenetic disorder first described by Angelman in 1965 as a condition marked by severe developmental delay, movement abnormalities, and a characteristic happy demeanor (1). It is primarily caused by genetic abnormalities involving chromosome 15q11–q13, including maternal deletions, mutations of the UBE3A gene, imprinting defects, and paternal uniparental disomy (2,14). The prevalence

of AS is estimated to range from 1 in 10,000 to 1 in 20,000 individuals (2,7).

Clinically, individuals with AS present with severe intellectual disability, minimal or absent speech, epilepsy, ataxia, and distinctive behavioral features such as frequent smiling, laughter, hyperexcitability, and hand-flapping movements (3,7,9). Neurological involvement is profound, with significant impairment in motor control, coordination, and postural stability, which contributes to delayed achievement of developmental milestones (5,14).

Motor dysfunction is a core feature of AS and has a major impact on functional independence. Children with AS exhibit delayed gross motor development, poor balance, abnormal gait patterns, and difficulties in transitional movements such as sitting, standing, and walking (10,11). Although some individuals achieve assisted or independent ambulation, motor skills commonly plateau at an early developmental stage, leading to long-term dependence in activities of daily living (11,14). Assessment of motor function and functional abilities in individuals with AS frequently utilizes standardized outcome measures such as the Gross Motor Function Measure (GMFM-66/88) and the Functional Independence Measure (FIM), which have demonstrated reliability in pediatric neurological populations (4,6). These tools provide objective evaluation of progress and guide therapeutic interventions.

Physiotherapy forms a fundamental component of multidisciplinary care in AS, with goals aimed at improving postural control, balance, gait, functional mobility, and preventing secondary musculoskeletal complications (12). Rehabilitation strategies often incorporate principles of motor control, task-oriented training, and neurodevelopmental facilitation to enhance motor learning and functional outcomes (5,15). Emerging evidence, including case reports and systematic reviews, suggests that structured motor rehabilitation programs such as robotic-assisted gait training combined with virtual and augmented reality may improve balance and gross motor function in children with AS (8,13,15). However, despite growing recognition of the importance of physiotherapy in AS management, the literature remains limited, with a scarcity of high-quality clinical trials. This highlights the need for further clinical documentation and case-based evidence to strengthen rehabilitation protocols and optimize functional outcomes for individuals with Angelman syndrome (13,15).

## **Aim**

To know the effect of a structured functional activity-based physiotherapy program with NDT approach on motor performance and functional independence in a child with Angelman syndrome.

## **Objective**

1. To determine the changes in gross motor abilities using GMFM before and after a 4-week physiotherapy intervention
2. To determine the changes in functional independence using WeeFIM before and after a 4-week physiotherapy intervention

## **CASE REPORT**

This prospective interventional case study was conducted after obtaining written informed consent from the parent. The patient was a 10-year-old female with Angelman syndrome, born of a non-consanguineous marriage at 34 weeks gestation via LSCS. She cried immediately after birth, required no NICU admission, and was discharged on day 7. At 3 months of age, she was hospitalized for pneumonia for 6–7 days. Delayed motor milestones were observed at 6 months, and neurological evaluation confirmed Angelman syndrome at 9 months of age.

Motor milestones achieved included rolling and supported sitting at approximately 3 years, independent sitting and creeping at 4–5 years, crawling at 7 years, and supported standing and walking at 8 years. At 10 years of age, the child demonstrated supported walking, pull-to-stand transitions, and limited short-distance mobility, requiring assistance for most functional activities.

## **Outcome Measures**

Outcome measures were administered at baseline and after 4 weeks of intervention. The Gross Motor Function Measure (GMFM) and Wee Functional Independence Measure (WeeFIM) were used to assess gross motor performance and functional independence.

### Gross Motor Function Measure (GMFM)(REF)

The Gross Motor Function Measure (GMFM-66 and GMFM-88) is a standardized observational assessment tool designed to evaluate gross motor function in children with neurological impairments. It assesses five dimensions: lying & rolling, sitting, crawling & kneeling, standing, and walking/running/jumping. Scores are expressed as a percentage, indicating the child's current gross motor performance relative to the maximum possible score. Higher scores indicate better motor function.

The GMFM is widely used in pediatric neurorehabilitation and has demonstrated high inter-rater and intra-rater reliability. It has strong construct validity and responsiveness to change, making it suitable for measuring intervention outcomes in children with developmental disorders.

### Wee Functional Independence Measure (WeeFIM) (REF)

The WeeFIM is a pediatric adaptation of the Functional Independence Measure (FIM)

used to assess functional independence in children aged 6 months to 7 years, and in older children with developmental disabilities. It evaluates performance across three domains: self-care, mobility, and cognition. Each item is scored on a 7-point scale ranging from total assistance (1) to complete independence (7). Higher scores indicate greater functional independence.

The WeeFIM has demonstrated good reliability and validity in pediatric neurological populations. It is sensitive to functional changes over time and is useful in evaluating the impact of rehabilitation interventions on daily living skills.

### Intervention

Assent was taken and ethical approval was taken for the case study.

The child received a structured functional activity-based physiotherapy program for 4 weeks, 5 sessions per week, with each session lasting approximately 45 minutes. The intervention incorporated Neurodevelopmental Treatment principles and emphasized task-oriented functional activities.

**Table 1: Physiotherapy Intervention Program**

Intervention Component	Activities	Purpose
Postural Control Training	Supported sitting, trunk facilitation, weight shifting	Improve trunk stability
Transition Training	Sit-to-stand, supine-to-sit with assistance	Enhance functional mobility
Balance Training	Static and dynamic standing with support	Improve balance and symmetry
Gait Training	Supported walking, step facilitation	Enhance ambulation

## RESULT

Post-intervention assessment demonstrated functional improvement in gross motor abilities and functional independence following the 4-week physiotherapy program.

**Table 2: Outcome Measures Pre- and Post-Intervention**

Outcome Measure	Pre-intervention	Post-intervention
GMFM (%)	54.46	60.82
WeeFIM	Lower level of assistance	Improved functional independence

## DISCUSSION

Angelman syndrome (AS) is a rare neurogenetic disorder associated with profound developmental delay, impaired motor coordination, balance deficits, epilepsy, and significant limitations in functional independence. Although

individuals with AS generally have a near-normal life expectancy, persistent motor dysfunction necessitates long-term rehabilitation and continuous caregiver support. Deficits in postural control, motor planning, and gait coordination markedly restrict participation in activities of daily

living and negatively affect quality of life (2). In the present case study, a structured 4-week physiotherapy program based on functional activity training and Neurodevelopmental Treatment (NDT) principles resulted in measurable improvements in gross motor performance and functional independence. The increase in GMFM score from 54.46% to 60.82% indicates enhanced postural stability, improved transitional movements, and better supported ambulation. These changes are clinically meaningful, as even modest improvements in gross motor abilities can translate into increased autonomy and reduced caregiver burden in children with severe neurodevelopmental disorders (4).

Motor development in Angelman syndrome is typically characterized by early plateauing, with most children achieving motor skills equivalent to 24–30 months of developmental age. Consequently, rehabilitation goals often focus on optimizing existing functional abilities, improving movement efficiency, and preventing secondary complications rather than achieving age-appropriate milestones. The functional gains observed in this 10-year-old child demonstrate that neuroplasticity and motor learning remain possible beyond early childhood when therapy is individualized, repetitive, and task-specific (4). This finding highlights the importance of continued rehabilitation even in older children with AS.

Neurodevelopmental Treatment principles emphasize facilitation of normal movement patterns, postural alignment, and controlled weight shifting during functional tasks. In this case, repeated practice of sit-to-stand transitions, supported standing, balance activities, and guided gait training likely contributed to improved symmetry in weight bearing, enhanced trunk control, and improved mobility. Similar benefits of task-oriented and NDT-based interventions have been reported in children with other neurodevelopmental disorders, supporting the applicability of these principles in

Angelman syndrome despite etiological differences (6).

Functional independence is a major concern for families of children with Angelman syndrome. Improvements in WeeFIM scores observed in this study reflect enhanced participation in mobility and transfer-related activities. Previous research indicates that even limited gains in functional mobility can significantly reduce caregiver workload, improve ease of handling, and enhance family quality of life, underscoring the clinical relevance of functional outcome measures in pediatric rehabilitation (5). Although alternative therapies such as hydrotherapy, hippotherapy, and music therapy have been reported to improve engagement and psychosocial well-being in children with AS, robust scientific evidence supporting their effectiveness in improving motor outcomes remains limited. Additionally, heterogeneity in genetic subtypes, severity of impairment, and intervention protocols across studies restrict meaningful comparison of outcomes (7). Therefore, physiotherapy programs should be individualized, goal-oriented, and focused on meaningful functional activities relevant to daily life.

This case study is limited by its single-subject design and short intervention duration, which restrict generalizability of findings. Nevertheless, it provides valuable clinical evidence supporting the role of functional activity-based physiotherapy in improving gross motor performance and functional independence in children with Angelman syndrome. Future studies involving larger samples and longer follow-up periods are required to establish standardized rehabilitation protocols and evaluate long-term outcomes.

## **CONCLUSION**

This case study demonstrates that a structured, goal-oriented functional activity-based physiotherapy program incorporating Neurodevelopmental Treatment (NDT) can result in measurable improvements in gross motor performance and functional

independence in a child with Angelman syndrome. The improvement in GMFM scores and enhanced WeeFIM performance indicate that even a short-duration, intensive rehabilitation program can facilitate motor learning, improve postural control, and enhance mobility. Although motor development in Angelman syndrome typically plateaus early, this study supports the concept that neuroplastic changes and functional gains remain possible with individualized, repetitive, and task-specific interventions.

### **Clinical Implications**

- Early and continuous physiotherapy intervention is essential to optimize motor potential in children with Angelman syndrome.
- Functional activity-based training enhances carryover into activities of daily living.
- Even modest improvements in GMFM scores may significantly reduce caregiver burden.
- Regular use of standardized outcome measures such as GMFM and WeeFIM allows objective monitoring of progress and treatment effectiveness.

### **Declaration by Authors**

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