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Overcoming Early Childhood Developmental Plateaus in 15q11.2 Deletion: A 4-Year Rehab Case Report

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INTRODUCTION

•Angelman syndrome (AS) is a rare neurogenetic disorder caused by loss of maternal UBE3A expression and is characterized by severe developmental delay, minimal speech, gait ataxia, and intellectual disability. Children with deletion-type AS generally exhibit more pronounced motor and communication impairments and frequently experience developmental plateaus in early childhood, limiting functional independence and school readiness. Although early intervention is widely recommended, fragmented care may reduce therapeutic consistency. This case study highlights how four years of uninterrupted multidisciplinary day-hospital rehabilitation helped overcome developmental plateaus in early childhood and restore developmental progress.

Case presentation

•A female child born at 34 weeks' gestation (2,290 g) was diagnosed with 15q11.2 deletion AS at 6 months of age. From November 2022 to February 2026, she participated in seven intensive day-hospital rehabilitation programs at our center (approximately 1,100 treatment days), ensuring continuous multidisciplinary management.

•Serial Gross Motor Function Measure (GMFM) assessments guided phase-specific physical therapy. Early intervention addressed delayed gait initiation and instability (GMFM 73.94%, Domain E: 36.1%). As functional standing and ambulation improved, therapeutic focus transitioned to pelvic control, dynamic balance, and spinal alignment management, including scoliosis prevention. These adjustments were associated with progressive improvement to GMFM 93% (Domain E: 77.7%) by February 2026, reflecting substantial gains in higher-level motor function.

•Occupational therapy advanced from basic grasp patterns to complex hand manipulation and activities of daily living (ADL) training, including utensil use and personal item management. Her Functional Independence Measure for Children (WeeFIM) score increased from 21 to 34, indicating reduced caregiver assistance and improved functional autonomy.

•Speech therapy was delivered intensively (3–5 sessions per week), progressing from vocalization induction to receptive and expressive communication training. Augmentative and alternative communication (AAC) strategies were introduced to address expressive limitations, enabling purposeful social interaction.

•Based on these cumulative functional improvements, the patient is scheduled to enter a special education school in March 2026.

Date	Total score	A	B	C	D	E
2022.11	73.94	73.8	66.6	36.1	100	98.3
2023.03	81.58	78.5	79.4	50.0	100	100
2023.10	87.68	95.2	79.4	63.8	100	100
2024.01	91.7	95.2	89.7	73.6	100	100
2024.10	92.04	92.8	89.7	77.7	100	100
2025.06	93.0	97.6	89.7	77.7	100	100
2026.02	93.0	97.6	89.7	77.7	100	100

Table 1. GMFM progression

CONCLUSION

•This four-year follow-up suggests that developmental plateaus in deletion-type AS may be modifiable through sustained, coordinated rehabilitation. Continuous single-center care allowed therapy goals to evolve alongside functional progress, resulting in meaningful gains across motor, ADL, and communication domains. Longitudinal multidisciplinary rehabilitation may facilitate successful transition from medical care to educational environments in children with severe neurogenetic disorders.

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