LONGITUDINAL CASE REPORT OF A FEMALE PATIENT WITH ANGELMAN SYNDROME

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INTRODUCTION

Angelman Syndrome (AS) is a neurodevelopmental disorder first described by Harry Angelman in 1965, with an estimated prevalence of 1 in 12,000–20,000 live births. It is most commonly caused by a deletion or mutation of the maternally inherited UBE3A gene on chromosome 15q11–q13. Clinical features include severe intellectual disability, absent or minimal speech, ataxic gait, seizures, and a characteristic happy demeanor. Motor impairments in AS are profound and persistent, often limiting independence into adulthood. Hypotonia, poor coordination, and balance deficits are common, and many individuals require lifelong mobility support. Physiotherapy is a key component of multidisciplinary care, aiming to maximize functional potential, prevent secondary complications, and enhance quality of life.

AIM

The purpose of this report is to present a case of a girl with Angelman syndrome and the development of motor skills over a period of 9 years.

CASE PRESENTATION

Patient is a female, diagnosed with AS (at age 6) via genetic testing confirming a maternal deletion in 15q11–q13. Condition at the start of the rehabilitation at age 2: Unable to stand without support, severe hypotonia, poor head and trunk control, frequent seizures (controlled with medications), sleep disturbances. Comorbidities: Epilepsy, mild scoliosis, gastroesophageal reflux. The patient lived with her parents, who were actively involved in her care and home exercise program. Rehabilitation Goals: Improve gross motor skills, enhance postural stability, promote independent mobility, prevent contractures and secondary musculoskeletal complications, facilitate participation in play and school activities. Therapy was delivered 3–5 sessions/week in early years, tapering to 2–3 sessions/week in later years, supplemented by a structured home exercise program.

RESULTS

Motor Milestones Age 3: Independent sitting, Age 4.5: Crawling and pulling to stand, Age 6: Independent walking with ankle foot orthoses, Age 9: Stair climbing with supervision

DISCUSSION

This case demonstrates that long term, structured physiotherapy can lead to substantial functional gains in AS. The patient achieved independent ambulation, improved endurance, and greater participation in daily activities. Previous studies report that many individuals with AS remain non ambulatory or require significant support. This case suggests that with sustained, intensive therapy, higher functional levels are achievable.

Measure	Baseline	Year 4	Year 8
GMFM-88 (%)	28	54	72
PEDI Mobility (scaled)	23	45	61
TUG (sec)	Unable	28	17
6MWT (m)	Unable	180	480

Table 2. Functional Outcomes Over Time

CONCLUSION

An 9 year physiotherapy program in a child with Angelman Syndrome resulted in marked improvements in gross motor function, mobility, and participation. This supports the integration of lifelong physiotherapy into standard AS care, with emphasis on individualized, goal oriented interventions.