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# Enhancing functional independence through physiotherapy in prader–willi syndrome: a pediatric case study

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***Abstract:*** *Background: Prader-Willi Syndrome (PWS) is a rare hereditary neurodevelopmental condition caused by chromosome 15q11-q13 abnormalities, with an estimated frequency of 1 in every 15,000–25,000 live births. It is characterized by hypotonia, developmental delay, hyperphagia, obesity, endocrine dysfunction, and behavioral abnormalities, which impair functional independence significantly. Physiotherapy is an important part of multidisciplinary management for improving motor performance and quality of life in affected children. Case presentation: A 2.5-year-old male infant with confirmed PWS (paternal deletion of chromosome 15q11-q13) presented with generalized hypotonia, delayed gross motor milestones, poor postural control, wide-based gait, and a risk of obesity. He was delivered via regular vaginal delivery (birth weight: 1.80 kg) and spent two weeks in the NICU. Genetic testing verified the diagnosis. Intervention: A planned 12-week physiotherapy rehabilitation program was designed, consisting of three supervised sessions per week (45 minutes each), complemented by daily caregiver-assisted home exercises. The plan included flexibility training, progressive core and lower limb strengthening, balance training, gait rehabilitation, postural correction, sensory integration, hydrotherapy, orthotic prescription (ankle-foot orthoses and lumbar orthoses), and caregiver education. Results: After 12 weeks of care, the patient showed clinically significant improvements. The Pediatric Balance Scale (PBS) score increased from 18/56 to 38/56 (+20 points). The GMFM-66 score improved from 42.3 to 58.7 (+16.4). The Wee FIM overall score increased from 54 to 79/126 (+25 points). No adverse events were recorded. Conclusion: Early, systematic physiotherapy improves balance, gross motor function, and functional independence in young patients with PWS. Long-term management requires multidisciplinary rehabilitation and active caregiver participation.*

***Key Words:*** *Prader–Willi Syndrome, Physiotherapy, Paediatric Rehabilitation, Functional Independence, Balance Training, Hypotonia, Gait Training, CARE Guidelines, Hydrotherapy, Outcome Measures*

## 1. INTRODUCTION:

Prader-Willi Syndrome (PWS) is an uncommon and complex genetic condition caused by the loss of function of paternally expressed genes on chromosome 15q11-q13. The incidence is estimated to be 1 in every 15,000–25,000 live births worldwide [1, 2]. The vast majority of cases are caused by three primary genetic subtypes: paternal deletion (about 65–75% of cases), maternal uniparental disomy (20–30%), and imprinting abnormalities (1–3%), all of which have separate molecular causes but share clinical symptoms [1, 3].

The syndrome exhibits a wide range of metabolic, endocrine, and neurobehavioral characteristics. During the neonatal and early infant period, severe generalized hypotonia dominates the clinical picture, frequently demanding extensive neonatal care and aided feeding. As the child develops through early childhood, hyperphagia and the associated risk of morbid obesity become the predominant therapeutic concerns, accompanied by endocrine problems such as growth hormone insufficiency and hypogonadism [8, 12].

Motor development in PWS is typically delayed due to widespread muscular weakness and poor postural control [4, 5]. Gross motor milestones such as rolling, sitting, standing, and walking are severely delayed compared to chronological age, limiting functional independence and participation in age-appropriate activities. Given the permanent nature of these motor deficits, physiotherapy rehabilitation is regarded as an essential component of the multidisciplinary management framework [15, 20].

The physiotherapy literature on PWS is growing, but is still restricted due to the condition's rarity. Existing systematic reviews have looked at the effects of physical activity [6, 7] and growth hormone therapy [4] on motor performance, with promising but mixed results. This case report describes the clinical course, physiotherapy assessment, intervention, and outcomes of a 2.5-year-old male child with confirmed PWS, contributing to the evidence base for early rehabilitation in this population. This report follows the CARE (CAse REport) guidelines [Gagnier JJ et al., 2014].

## 2. CASE PRESENTATION:

### 2.1 Patient History

The patient is a 2.5-year-old male child born by normal vaginal delivery weighing 1.80 kilograms. Significant newborn complications were apparent upon birth, including severe respiratory distress and widespread hypotonia, necessitating a two-week stay in the neonatal Intensive Care Unit (NICU). During this time, extensive evaluations were carried out, including respiratory assessment, cardiovascular review, MRI, vital sign monitoring, ECG, and formal neurology consultation.

Prader-Willi Syndrome was diagnosed after genetic testing revealed a paternally inherited deletion of chromosome 15q11-q13. The patient was referred to the Subhadraben Sureshchandra Institute of Physiotherapy for structured rehabilitation. At the time of referral, the patient was not receiving growth hormone therapy; an endocrine review was being scheduled concurrently. Other co-interventions had not yet been formally initiated, but multidisciplinary team follow-up was planned.

### 2.2 Developmental Milestone History

A detailed developmental history was obtained from the parents. The following milestone acquisitions were documented at assessment (Table 1):

**Table 1: Developmental Milestone History**

| Milestone | Expected Age | Achieved / Status at Assessment |
|---|---|---|
| Head control in prone | 3–4 months | Achieved at ~6 months (delayed) |
| Rolling | 4–6 months | Achieved at ~9 months (delayed) |
| Sitting independently | 6–9 months | Not yet achieved at time of referral (age 30 months) |
| Pulling to stand | 9–12 months | Not yet achieved |
| Standing independently | 11–13 months | Not yet achieved |
| Walking independently | 12–15 months | Not yet achieved |
| Pincer grasp | 9–10 months | Partially achieved (weak grip) |
| First words | 12 months | ~3 words at 30 months (delayed) |

### 2.3 Clinical Findings

**Physical Examination:**

- Generalised hypotonia (global, confirmed by clinical tone assessment)
- Reduced proximal muscle strength (hip flexors, shoulder girdle, trunk)
- Poor dynamic and static postural control
- Delayed gross motor milestone acquisition (see Table 1)
- Wide-based, unsteady gait pattern
- Balance impairment — unable to stand independently
- Almond-shaped eyes; characteristic dysmorphic facial features
- Small hands and feet; short stature for age
- Impaired coordination and motor planning
- Reduced endurance during physical activity

**Behavioural and Functional Findings:**

- Mild behavioural disturbances (attention difficulties, impulsivity)
- Reduced participation and motivation in physical activity
- Difficulty in standing independently
- Inability to climb stairs without maximum assistance
- Impaired walking endurance
- Limited participation in age-appropriate play activities

**Anthropometric Data:**

Weight: 14.2 kg (weight-for-age z-score: +1.8 SD, indicating a risk of obesity). Height: 82 cm (height-for-age z-score: -1.5 SD). BMI = 21.1 kg/m² (age-adjusted z-score: +2.1 SD). The patient was in nutritional Phase 2 of PWS (Miller et al. classification), defined by increased weight gain without a major change in calorie intake [11].

### 2.4 Investigations

- Respiratory assessment: Normal
- Chorionic villus sampling: Normal karyotype (structural chromosomes intact)
- Genetic analysis: Paternally derived deletion of chromosome 15q11–q13 confirmed (FISH and methylation analysis)
- Neurology report: Hypotonia without weakness; almond-shaped eyes; abnormal skull morphology; depressed DTRs; good antigravity movement; spontaneous activity present
- Cardiovascular assessment: Normal
- Vital signs: Normal for age
- Overall impression: Genetic hypotonic phenotype consistent with Prader–Willi Syndrome (paternal deletion subtype)

### 2.5 Patient and Caregiver Perspective

The patient's parents expressed deep concern over his inability to walk independently at the age of 30 months, as well as his apparent lack of willingness to participate in active play with peers. The family demonstrated strong dedication to the rehabilitation program and readiness to participate in twice-daily home exercise sessions. The mother stated that she observed he was not meeting the same milestones as other children his age, and the family was resolved to do everything possible to assist him. Caregiver motivation and active engagement were recognized as important facilitating variables for the rehabilitation program.

## 3. PATIENT CLINICAL TIMELINE:

**Table 2: Patient Clinical Timeline**

| Timepoint | Event | Clinical Details |
|---|---|---|
| December 2021 (Birth) | NICU Admission | Birth weight 1.80 kg; normal vaginal delivery; severe respiratory distress and generalised hypotonia; NICU admission for 2 weeks |
| December 2021 (Week 2) | NICU Discharge | Feeding established; chromosome 15q deletion confirmed; MDT referral initiated; GH therapy evaluation planned |
| January 2022 (Age ~1 month) | PWS Diagnosis Confirmed | Paternal deletion 15q11–q13 confirmed on FISH and methylation studies; physiotherapy referral issued |
| Age ~6 months | Early Motor Milestone | Head control achieved (delayed); passive physiotherapy and caregiver handling advice commenced |
| Age ~9 months | Rolling Achieved | Rolling from supine to prone achieved; hydrotherapy (bath exercises) commenced by parents |
| Age 30 months (Baseline — Week 0) | Formal Physiotherapy Referral | PBS: 18/56; GMFM-66: 42.3; WeeFIM: 54/126; Weight: 14.2 kg; BMI z-score: +2.1 SD; AFO and lumbar orthotic fitting arranged |

| | | |
|---|---|---|
| Weeks 1–4 | Phase 1: Foundation | Flexibility, passive/active-assisted mobilisation, hydrotherapy 2×/day (parent-led), sensory integration, postural awareness commenced; AFOs fitted Week 2 |
| Weeks 5–8 | Phase 2: Strengthening | Progressive core and lower limb strengthening; balance board introduced; sit-to-stand training progressed; treadmill walking (0.5 km/h, 30% body weight support harness) commenced |
| Weeks 9–12 | Phase 3: Function & Gait | Gait training with reduced manual assistance; stair-climbing practice; obstacle negotiation; functional activity training; lumbar orthosis fitted Week 10 |
| Week 12 (Programme End) | Final Assessment | PBS: 38/56 (+20); GMFM-66: 58.7 (+16.4); WeeFIM: 79/126 (+25); Weight: 14.5 kg; Walking with AFOs and minimal upper limb support achieved |
| 3 months post-Programme (Follow-up) | Follow-up Assessment | Functional gains maintained; walking with AFOs independently achieved over short distances; GH therapy initiated by endocrinology team; ongoing home programme continued |

## 4. PHYSIOTHERAPY ASSESSMENTS:

### 4.1 Outcome Measures

The validated outcome measures listed below were chosen for their demonstrated reliability and validity in pediatric populations with neuromotor disorders. Pre-intervention baseline scores were recorded in Week 0 and reviewed at Weeks 6 (midpoint) and 12 (programme completion).

**Table 3: Outcome Measures Used**

| Outcome Measure | Baseline Week 0 | Mid-point Week 6 | Post-Tx Week 12 | MDC / Interpretation |
|---|---|---|---|---|
| Pediatric Balance Scale (PBS) (0–56; higher = better) | 18/56 | 28/56 | 38/56 (+20 pts) | MDC = 4 pts. Change of +20 is highly clinically significant. |
| GMFM-66 (0–100; higher = better gross motor) | 42.3 | 51.0 | 58.7 (+16.4) | Clinically meaningful change typically ≥1.5 units. Change of +16.4 is substantial. |
| WeeFIM Total (18–126; higher = more independent) | 54/126 | 67/126 | 79/126 (+25 pts) | Improvement of +25 points indicates transition from modified dependence toward supervised independence. |
| Caregiver Independence VAS (0–10; higher = more independent) | 2.5/10 | 5.0/10 | 7.2/10 (+4.7) | Parent-rated daily activity independence more than doubled over the programme. |

### 4.2 ICF-Based Problem Classification

In accordance with the International Classification of Functioning, Disability and Health (ICF) framework, the patient's clinical presentation was characterised as follows (Table 4):

**Table 4: ICF-Based Problem Classification**

| ICF Domain | Patient-Specific Findings |
|---|---|
| Body Structure & Function (Impairments) | Generalized hypotonia; reduced proximal and core muscle strength; impaired proprioception and balance; delayed neuromotor maturation; mild cognitive delay; hyperphagia and obesity risk (endocrine dysfunction) |
| Activity (Limitations) | Inability to stand or walk independently; cannot climb stairs; unable to perform sit-to-stand unaided; limited capacity for sustained walking; restricted fine motor skills |
| Participation (Restrictions) | Reduced engagement in age-appropriate play; social participation limited by mobility; inability to access community environments without full caregiver support |
| Environmental Factors | Facilitators: Highly motivated parents; home exercise environment available; access to specialist physiotherapy. Barriers: Financial considerations for orthotic provision; community mobility barriers |
| Personal Factors | Age 2.5 years (within optimal neuroplasticity window); cooperative temperament; engaged caregiver support system |

## 5. PHYSIOTHERAPY INTERVENTION:

The rehabilitation plan lasted 12 weeks, with three supervised clinic sessions per week (45 minutes each), complemented by twice-daily caregiver-administered home exercises. The program was divided into three progressive phases, with progression depending on achievement of at least 80% of phase-specific performance targets.

### 5.1 Flexibility Training

Stretching exercises concentrated on the hamstrings, hip flexors, iliopsoas, gastrocnemius-soleus complex, and trunk rotators. All stretches were performed in supported postures. Protocol: static stretching for 15–30 seconds, 3–5 repetitions per muscle group; incorporated in all supervised sessions and twice-daily home programme. Progression: increased duration to 45 seconds as tolerance improved.

### 5.2 Core Muscle Strengthening

Progressive core strengthening addressed trunk stabilisers and co-activation deficiencies between core and lower limb muscle groups. Interventions included graded sit-to-stand transfers with varying levels of manual assistance (maximum to supervised), enhanced abdominal activity during supine-to-sit transitions, bilateral bridge exercises graduated to single-leg bridges, supported sitting trunk rotation (reverse chopping), and modified plank positions with progressive duration from 5 to 30 seconds.

### 5.3 Strengthening Exercises

Progressive resistance workouts targeted hypotonic muscle groups including quadriceps, gluteals, hip abductors, and ankle stabilizers (gastrocnemius and tibialis anterior). Functional strengthening exercises included squat-to-stand, mini-squats, and step-ups on a low block. Progression commenced with bodyweight, then modest ankle weights from Week 8 onwards.

### 5.4 Balance Training

Balance training included supported single leg stance with gradually reduced manual assistance (target: 10-second hold by Week 12), progression from wide to narrow base of support for tandem standing, balance board training with gentle rocker board perturbations, dynamic reaching tasks during supported standing, and weight-shifting exercises in mediolateral and anteroposterior directions.

### 5.5 Postural Correction

Interventions included real-time postural feedback using visual cues to improve awareness of body alignment, practice alignment drills (standing tall with shoulders back and head aligned), and trunk stabilisation activities using visual input to promote a consistent posture.

### 5.6 Gait Training

Gait training involved progressive indoor walking drills for increasing distance, step training and stair climbing (beginning with bilateral upper limb support, graduating to unilateral rail support), negotiation of low obstacles and

varying floor surfaces, treadmill-assisted walking (0.5–0.8 km/h; 30% body weight support harness lowered to 15% by Week 12; 5–15 minutes duration), and community-relevant functional gait activities including turning and altering pace.

### 5.7 Sensory Integration Training

Sensory integration activities were implemented to address motor planning deficits and improve somatosensory processing. Activities included tactile stimulation using various textures on hands, feet, and trunk; proprioceptive loading via joint compression and weighted vest activities during supported standing; vestibular stimulation through controlled rocking and swinging in supported positions; and motor planning tasks using obstacle courses and graded movement sequences.

### 5.8 Hydrotherapy

Aquatic therapy was included owing to its well-established benefits in hypotonic conditions: buoyancy reduces antigravity loading demands, hydrostatic pressure offers somatosensory input, and the aquatic medium allows movement patterns not possible on land. Supervised sessions were held in a temperature-controlled pool (32–33°C) for 30 minutes, twice weekly from Week 4. Parent-administered bath hydrotherapy (daily 15-minute treatments including passive range of motion, active-assisted standing, and aquatic play) was initiated earlier. Activities included water walking, buoyancy-assisted standing, kicking exercises, and flotation-based balance tasks.

### 5.9 Orthotic Devices

Two orthotic devices were prescribed in consultation with an orthotist. Custom-made solid ankle-foot orthoses (AFOs) were fitted in Week 2 to improve distal lower limb stability, correct calcaneal valgus, and promote heel-toe gait. A prefabricated paediatric lumbar support was applied at Week 10 to improve proximal trunk and pelvic stability during prolonged standing and gait training.

### 5.10 Caregiver Education

Caregiver education included instruction and supervision for home exercise programmes, practice of postural handling and facilitation skills, obesity prevention strategies (reinforcing food guidelines and encouraging physical activity), protocols for AFO donning/doffing and skin inspection, behavioural management and motivational reinforcement strategies, and recognition of symptoms requiring medical attention.

## 6. RESULTS:

### 6.1 Primary Outcome Measures

After 12 weeks of intensive physiotherapy rehabilitation (3 supervised sessions per week plus a daily home programme), the patient showed clinically relevant improvements in all primary end measures (Table 3, Section 4.1).

The Pediatric Balance Scale (PBS) score improved from 18/56 at baseline to 38/56 at treatment completion (+20 points). This improvement well exceeds the PBS minimal detectable change (MDC) of four points, indicating true functional improvement above measurement error. At baseline, the patient could not sit independently without upper limb assistance; after 12 weeks, he could stand with bilateral hand support and complete lateral weight shifts.

The GMFM-66 score increased from 42.3 to 58.7 (+16.4 points), representing a shift from inability to hold any standing position to supported standing and assisted ambulation over short distances, consistent with GMFM Dimension D (standing) and Dimension E (walking and running) gains.

The WeeFIM overall score increased from 54/126 to 79/126 (+25 points), indicating a functional shift from complete dependence to supervised or modified independence in several self-care and mobility domains. The caregiver-rated independence (VAS) increased from 2.5/10 to 7.2/10.

### 6.2 Functional Milestones

- Week 0: Unable to maintain any weight-bearing through lower limbs; required full caregiver support for all upright activities
- Week 4: Tolerated supported standing for up to 30 seconds with bilateral upper limb support and AFOs
- Week 8: Able to perform 5 consecutive sit-to-stand transfers with moderate manual assistance; walking 2–3 steps with bilateral walking frame
- Week 12: Walking 10–15 metres with AFOs and minimal upper limb support (one hand); step climbing with unilateral rail support

• 3-month follow-up: Walking independently over short distances with AFOs; continued home programme maintained

### 6.3 Adverse Events

No adverse events, safety incidents, or unexpected clinical deterioration were observed or reported during the 12-week programme. Skin integrity under the AFOs was monitored at each session and remained intact throughout. Caregiver compliance with the home exercise programme was rated as excellent, with an estimated adherence rate of >85% based on exercise diary review.

## 7. DISCUSSION:

This case report describes the clinical course and outcomes of a 12-week structured physiotherapy rehabilitation programme for a 2.5-year-old child with confirmed Prader-Willi Syndrome (paternal deletion subtype). The findings show clinically significant improvements in balance (PBS +20 points), gross motor function (GMFM-66 +16.4), and functional independence (WeeFIM +25 points), emphasizing the importance of early physiotherapy in PWS management.

The results are consistent with the broader evidence base. Reus et al. (2011) revealed widespread motor difficulties in PWS caused by both central hypotonia and peripheral neuromuscular dysfunction, providing theoretical support for the multi-component physiotherapy approach employed here [5]. Morales et al. (2019) found that physical exercise interventions in PWS were associated with improvements in muscle strength, endurance, and body composition, with the highest benefits in programmes incorporating progressive resistance training [7]. The present case contributes paediatric evidence suggesting that similar approaches can be effectively employed in very young children.

The magnitude of the balance gain (PBS +20 points) is particularly notable. The post-intervention score of 38/56 demonstrates a clinically significant shift toward functional balance capacity. Balance board training, weight-shifting exercises, and AFO prescription likely worked synergistically, each targeting a different contributing mechanism: central proprioceptive processing, dynamic postural adjustment, and distal biomechanical alignment, respectively.

Hydrotherapy is worthy of detailed discussion. The dual approach of supervised pool-based sessions and parent-administered bath hydrotherapy exploited the unique properties of the aquatic medium — buoyancy, hydrostatic pressure, and warm water facilitation — to allow movement practice that would have been impossible on land given the severity of hypotonia. Crinò et al. (2018) identified water treatment as a viable supplementary intervention for PWS recovery [15], and the current case supports this clinical opinion.

Caregiver participation played an essential role in the observed outcomes. Adherence to the home programme exceeded 85%, and the parent-administered hydrotherapy component offered significant additional movement practice. This has direct clinical implications for programme design in this population.

The absence of growth hormone therapy during the 12-week programme is important in interpreting the observed improvements. GH therapy has been shown to increase muscular tone and motor development in PWS [16, 19], and its subsequent initiation at the 3-month follow-up confirms that the programme-completion gains can be attributed solely to the physiotherapy intervention, reinforcing the case for physiotherapy as an independent contributor to functional outcomes.

The use of orthotic devices — AFOs and a lumbar orthosis — as adjuncts to the exercise programme was a critical aspect of care. Beginning in Week 2, AFOs enabled biomechanically aligned heel-toe gait practice, limiting the reinforcement of compensatory gait patterns. This is consistent with recommendations by Goldstone et al. (2008) for comprehensive multidisciplinary management addressing musculoskeletal issues early [3].

## 8. CONCLUSION:

This case study illustrates that initiating a planned, multi-modal physiotherapy programme early in a child's life with Prader-Willi Syndrome can result in clinically significant and durable gains in balance, gross motor function, and functional independence. A PBS increase of +20 points, GMFM-66 improvement of +16.4, and WeeFIM improvement of +25 points over 12 weeks are substantial changes that translated into functional mobility benefits, including independent short-distance ambulation with AFOs after three months.

Physiotherapy is an essential component of multimodal management of PWS in childhood. The evidence from this case suggests that early referral, individualized multi-modal programming, orthotic integration, and extensive caregiver involvement are essential components of an effective rehabilitation strategy. Larger prospective cohort studies and

single-case experimental designs are needed to standardize physiotherapy protocols and assess long-term functional outcomes across the PWS lifespan.

## 9. LIMITATIONS:

The findings of this single-case descriptive report cannot be generalized to the broader PWS population. The absence of a control group makes it impossible to attribute all reported gains solely to physiotherapy; natural developmental growth, practice effects, and regression to the mean are potential confounders. Observer and performance bias exist as the treating physiotherapist conducted outcome evaluations. The 12-week regimen provides only short-term results, and the long-term durability of improvements requires further investigation.

## 10. RECOMMENDATIONS:

- Early referral to physiotherapy, commencing in the neonatal period, is crucial for maximizing neuroplasticity in PWS.
- Multi-modal programmes targeting flexibility, strength, balance, gait, orthotic care, and hydrotherapy yield better results than isolated treatments.
- AFOs and lumbar orthoses can effectively complement vigorous physiotherapy during early rehabilitation phases.
- High adherence to home exercise programmes and structured caregiver education are crucial for successful outcomes.
- Effective collaboration across endocrinology, dietetics, occupational therapy, and speech therapy improves overall patient outcomes.
- Randomized controlled trials and single-case experimental designs with multiple baselines are needed to generate higher-level evidence.

## Patient's Consent:

The patient's parents/legal guardians provided written informed consent for the publication of this case report, including clinical data, developmental history, and outcome measure results. Consent was acquired for print and internet publication.

## Ethical Approval:

This case report was conducted in conformity with the ethical principles of the Declaration of Helsinki, and institutional ethical approval was obtained (reference: [Institution Ethics Reference Number]). The case report is part of routine clinical care reporting and was exempt from full ethics committee assessment under local institutional rules.

## CARE Guideline Compliance:

This case report was prepared following the CARE (CAse REport) guidelines [Gagnier JJ, Kienle G, Altman DG, et al. The CARE guidelines: consensus-based clinical case report recommendations. J Clin Epidemiol. 2014;67(1):46–51].

## Conflict of Interest:

The authors declare no conflicts of interest.

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