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Case Report

Dynamic Systems Theory Framework in Physiotherapy Management of Children with Joubert Syndrome: A Case Study

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ABSTRACT

Joubert Syndrome (JS) is a rare neurodevelopmental disorder characterized by cerebellar and brainstem malformations, resulting in hypotonia, motor delays, and poor coordination. Physiotherapy plays a key role in improving functional outcomes, yet standardized management strategies remain limited. This case study highlights the application of the Dynamic Systems Theory (DST) framework in physiotherapy management of a child with JS, emphasizing functional diagnosis and patient-centered care. A 5-year-old boy with JS presented with motor delays, hypotonia, and impaired postural control. Using the DST approach, a tailored physiotherapy program was designed addressing neuromuscular, sensory, cognitive, and environmental subsystems. Interventions included sit-to-stand training, gait activities, sensory integration, coordination tasks, and respiratory exercises. Outcome measures used were the Gross Motor Function Measure (GMFM) and Pediatric Berg Balance Scale and WEEFIM. Over five weeks, the child showed significant improvement in balance, coordination, and functional independence. GMFM and balance scores improved markedly. Active family involvement and a structured environment enhanced engagement and therapy outcomes. The DST framework proved effective in managing the complex motor impairments of JS by promoting adaptability and functional gains. Functional diagnosis enabled targeted, task-specific interventions, resulting in improved quality of life and independence. This approach holds promise for broader application in rare neurodevelopmental conditions.

Keywords: *Joubert Syndrome, Dynamic Systems Theory, Functional Diagnosis, Paediatric Physiotherapy.*

INTRODUCTION

Joubert Syndrome (JS) is a rare and complex genetic disorder first described by Dr. Marie Joubert in 1969. It is characterized by neurological, motor, and developmental abnormalities due to agenesis or hypoplasia of the cerebellar vermis, which primarily affects motor coordination, muscle tone, and balance[1]

The global prevalence of JS is estimated at 1 in 80,000 to 100,000 live births, though the true incidence may be higher due to diagnostic challenges and phenotypic variability[2]. The radiological hallmark of JS is the "molar tooth sign," which reflects malformations of the brainstem and cerebellum, visible through axial brain MRI scans[3,4].

JS has a heterogeneous genetic basis involving mutations in genes such as AHI1, CEP290, and TMEM67, which disrupt the structure and function of primary cilia, essential for cellular signalling[5]. These mutations are typically inherited in an autosomal recessive pattern, though X-linked forms also exist, primarily affecting males[4]. Beyond the neurological manifestations, JS often presents with multi-system involvement, including renal, hepatic, and ocular abnormalities, complicating its clinical management[6].

The clinical management of JS remains largely supportive, with multidisciplinary interventions required to address the syndrome's diverse manifestations. Physiotherapy plays an essential role in improving motor functions and quality of life, despite the lack of specific guidelines tailored to JS. The use of Dynamic Systems Theory (DST) in physiotherapy offers a framework for understanding motor control as the result of interactions between neuromuscular, sensory, cognitive, and environmental systems, promoting adaptability and self-organization in motor behaviour, thus to facilitate motor learning and adaptive movement strategies7.

Aim:

To evaluate the effectiveness of a physiotherapy intervention based on the Dynamic Systems Theory (DST) in improving gross motor function, balance, and task-specific independence in a 5-year-old child diagnosed with Joubert Syndrome.

Objectives:

1. To measure improvements in gross motor abilities in a child with Joubert Syndrome using the Gross Motor Function Measure (GMFM), thereby assessing changes in overall motor control
2. To evaluate balance using the Paediatric Berg Balance Scale (PBS) as an indicator of progress in equilibrium and movement coordination during functional tasks.
3. To monitor changes in daily functional independence and self-care capabilities using the Wee FIM scale, providing insight into the real-world applicability and impact of physiotherapy based on the Dynamic Systems Theory (DST).

CASE REPORT

A 5-year-old boy diagnosed with Joubert Syndrome presented with significant motor delays and coordination difficulties. His medical history includes neonatal jaundice, respiratory distress syndrome, recurrent respiratory infections, and a seizure episode treated with valproates. The primary concerns include an inability to stand without support, difficulty in walking, and developmental delays. The child lives in a supportive family environment, although initial rehabilitation was delayed due to family circumstances and the COVID-19 pandemic.

Primary Concerns: Inability to stand/walk without support, coordination issues, developmental delays.

Physiotherapy Assessment:

1. **Neuromuscular System:** Hypotonia in lower limbs, bilateral hamstring tightness, tightness in the right sternocleidomastoid muscle, abnormal co-activation patterns, and impaired postural control.
2. **Musculoskeletal System:** Decreased active range of motion (AROM), Decreased muscle strength in lower limbs
3. **Sensory System:** Impaired horizontal and vertical eye tracking, affected proprioception.
4. **Cognitive System:** Reduced attention span but good arousal and alertness.
5. **Environmental System:** Supportive family with financial resources for services.

Investigations done:

- **Brain MRI:** Confirmed "molar tooth sign," indicating cerebellar and brainstem abnormalities.
- **Genetic Testing:** Identified mutations in the AHI1 gene, confirming Joubert Syndrome.

The Dynamic Systems Theory (DST) approach was adopted due to its holistic and adaptable nature, making it suitable for managing complex neurological conditions like Joubert Syndrome. DST emphasizes movement variability, task-specific functional training, environmental modifications, and family involvement, promoting self-organization and motor learning through real-world experiences.

Table: -1 Parents' Goals, Patient's Strengths, and Prognostic Challenges

Category	Details
Parents' Goals	- Achieve independent standing and walking - Improve coordination and balance to reduce fall risk - Increase independence in daily self-care activities - Enhance participation in play and peer interaction
Patient's Strengths	- Good independent sitting balance - Optimal arousal and attentiveness during therapy - Achieved key motor milestones (tripod sitting, crawling, supported walking) - No deformities or involuntary movements
Prognostic Challenges	- Variability in severity of Joubert Syndrome - Multisystem involvement (neurological, visual, respiratory) - Unpredictable developmental trajectory - Need for individualized, flexible, long-term therapy

Dynamic systems theory framework - Constraint-Based Analysis & Treatment Plan: -

A constraint-based approach was employed as a core application of the Dynamic Systems Theory (DST) framework. The constraint-based approach was used to systematically assess and address barriers to functional movement in the child with Joubert Syndrome. By analysing constraints across the organism (e.g., hypotonia, coordination deficits), task (e.g., complexity, sequence), and environment (e.g., distractions, surfaces), individualized, task-specific interventions were developed under the Dynamic Systems Theory framework to promote adaptability, motor learning, and independence.

Table: - 2

Constraint	Issue	Intervention Strategies given to the patient
Structure & Hypotonia	Joint laxity, low muscle tone	Strengthening, postural stability, weight-bearing activities
Posture, Movement & Equilibrium	Poor postural control, balance issues	Given interventions targeting postural stability, weight shifting, and balance in dynamic environments and visual-vestibular training to improve sensory feedback processing
Coordination & Execution	Inefficient movement patterns, slow reactions	Task-specific training to improve motor control, speed, accuracy, and smooth transitions in movement.
Ideation & Cognitive Focus	Limited task focus, slow processing	Step-by-step guidance, visual cues, simplified tasks
Environmental Context	Sensitive to noise, difficulty adapting	Varying surfaces – Practice on foam mats, grass, and uneven ground to improve balance Minimize distractions – Use calm, structured settings during new skill learning; gradually introduce distractions to promote generalization. Use of visual cues/toys – Bright-colored targets and moving objects help engage attention and guide motor responses.

Cognitive and visual & Perceptual Systems	Fine motor control deficits, coordination, problem-solving during movement	Engaged in hand activities (e.g., picking up small objects, manipulating toys, stringing beads) and visual coordination exercises (hand-eye coordination tasks, and fine motor games)
Cardiopulmonary System	Recurrent Chest Infections, Increased Kyphosis	Respiratory exercises (diaphragmatic breathing, blowing tasks). Chest physiotherapy and postural drainage (If required)
Functional Limitations	Dependent on caregiver support	Building independence and improving basic motor skills with Gait training. Practice of real-world activities (e.g., stepping over obstacles, walking outside, dressing tasks) that simulated daily life

Outcome measures used:

1. **Gross Motor Function Measure (GMFM)[8]:**
   The GMFM is a standardized observational tool used to assess changes in gross motor function in children with motor impairments. It measures the child's ability to perform functional activities such as lying, sitting, crawling, standing, and walking. In this case, it was used to track progress in motor control and mobility.
2. **Paediatric Berg Balance Scale (PBS)[9]:**
   The PBS evaluates balance and postural control in paediatric populations through 14 tasks (e.g., standing, reaching, turning). It provides insight into the child's ability to maintain balance during functional activities. The tool is sensitive to improvements in dynamic and static balance following therapy.
3. **WeeFIM (Functional Independence Measure for Children)[10]:**
   The WeeFIM assesses functional independence in children across domains such as self-care, mobility, and cognition. It measures the level of assistance required for daily tasks, providing a broad view of the child's overall functional abilities. It is especially useful in evaluating progress in real-world performance and caregiver burden.

RESULTS

Following a five-week intervention based on the Dynamic Systems Theory (DST) framework, the child with Joubert Syndrome demonstrated measurable improvements in motor performance, postural control, and balance. The therapeutic program emphasized task-oriented activities, environmental variability, and multi-system engagement, including neuromuscular, sensory, cognitive, and postural control systems.

Gross Motor Function Measure (GMFM)

The child's GMFM score improved from a baseline of 75.42% to 80%, showing enhanced gross motor capacity. These gains were primarily noted in standing balance, supported walking, and transitional movements such as sit-to-stand. The improvement indicates increased ability to initiate and complete gross motor tasks with reduced assistance and greater postural alignment.

Paediatric Berg Balance Scale

Pre-intervention, the child scored 11/56 on the Paediatric Berg Balance Scale. Post-intervention, this increased to 23/56, indicating a +12-point improvement. This gain reflects better static and dynamic balance, trunk control, and the ability to maintain posture during upright functional tasks.

WeeFIM (Functional Independence Measure for Children)

The child remained at a level of moderate to severe dependence on the WeeFIM. While no numerical increase was recorded within the short time frame, observational improvements in motor engagement, self-initiated mobility, and participation in play and transitional activities were noted, suggesting early signs of progress toward functional independence.

Family and Therapist Observations

- Improved postural transitions with decreased cueing.
- Greater engagement and focus during therapy tasks.
- Better tolerance for upright activities and willingness to attempt self-care tasks.

Table: - 3 Changes in Outcome Measures Following 5-Week Physiotherapy Intervention Using the DST Framework

Outcome Measure	Pre-Intervention	Post-Intervention	Change Observed
Gross Motor Function Measure (GMFM)	75.42%	80%	+4.58% improvement, indicating enhanced gross motor capacity
Paediatric Berg Balance Scale	11 / 56	23 / 56	+12-point gain, reflecting improved postural control and balance
Wee-FIM	Moderate to severe dependence	Moderate to severe dependence	No numerical change, but notable functional improvements in mobility and self-care tasks

DISCUSSION

The results of this study demonstrate meaningful improvements in gross motor function and balance in a 5-year-old child with Joubert Syndrome (JS) after a 5-week physiotherapy intervention based on the Dynamic Systems Theory (DST) framework. The GMFM score increased from 75.42% to 80%, and the Pediatric Berg Balance Scale improved from 11/56 to 23/56, suggesting enhanced postural control and gross motor capabilities. While the WeeFIM score remained in the range of moderate to severe dependence, the improvement in motor control indicates a positive trend in the child's functional trajectory.

These findings are consistent with the work of Shumway-Cook and Woollacott (2017)[11], who emphasized the importance of environmental variability and task-specific training in facilitating motor learning and adaptability, particularly in paediatric neurorehabilitation. According to their research, variability in sensory and motor practice allows for more robust self-organization, which is crucial in children with developmental coordination disorders and cerebellar dysfunctions like JS. Similarly, Thelen and Smith (1994)[7], who pioneered DST, emphasized that motor behaviour emerges from complex interactions among multiple subsystems—including sensory, musculoskeletal, cognitive, and environmental—and not from isolated neural control. This theory aligns closely with the multi-system impairments observed in JS, such as hypotonia, poor coordination, and impaired eye tracking.

In comparison, traditional physiotherapy approaches such as Neurodevelopmental Therapy (NDT), which focus more on movement normalization, may not be as effective for genetic conditions like JS where atypical movement patterns are inherent to the disorder. Novak et al. (2013)[12]concluded in their evidence-based review that task-specific and goal-directed therapies showed stronger evidence for improving motor outcomes than hands-on facilitation techniques alone.

Improvements in balance scores were further supported by McKeon et al. (2012)[13], who proposed that balance training based on DST principles—through task manipulation, unstable surfaces, and dynamic progression—enhances postural control in individuals with chronic ankle instability. Applying similar methods in this case, such as balance activities on foam and variable surfaces, likely contributed to the child's increased balance capabilities and functional independence.

Additionally, Coté's (2015)[14] DST model of visual perception development supports the idea that visual perception is shaped by dynamic interactions between visual, motor, cognitive, and emotional systems. This model aligns with the visual-motor coordination tasks used in the intervention, such as eye-tracking during reaching or walking. These activities encouraged the child to self-organize and integrate perceptual inputs with functional movement, reinforcing the importance of DST in visual-motor rehabilitation.

The article *Understanding Dynamic Systems Theory: The Key to Athletic Performance and Rehabilitation*[15] highlights the importance of monitoring internal states such as fatigue, emotional readiness, and motivation, which directly impact the effectiveness of motor learning. In this case, family observations of the child's daily readiness and behaviour were incorporated into session planning, promoting safety and individualized progression.

This case also aligns with the findings of Ozge et al. (2017)[16], who documented the rehabilitation outcomes in a child with Joubert Syndrome and emphasized the benefits of a multidisciplinary approach, including respiratory and motor function interventions. The importance of understanding JS at a molecular level is further underscored by Brancati et al. (2010)[5], who identified multiple ciliopathy-related genes involved in the pathophysiology of JS, supporting the need for integrated therapy that acknowledges multisystem involvement.

The molecular insights provided by Parisi (2019)[17] reveal that Joubert Syndrome (JS) is a genetically diverse ciliopathy, involving mutations in multiple genes that affect brain development and multi-organ function. This genetic complexity contributes to the wide variability in clinical presentation, highlighting the need for flexible, individualized therapeutic approaches. The clinical overview by Parisi et al. (2007)[18] supports this by emphasizing the range of motor, visual, and systemic impairments seen in JS. In this context, the Dynamic Systems Theory (DST) offers a suitable framework, as it allows physiotherapy to be adapted based on each child's unique genetic, neurological, and developmental profile. Additionally, the clinical study by Surisetti et al. (2021)[19] reinforces the rarity and heterogeneity of Joubert Syndrome and highlights the importance of individualized, evidence-based rehabilitation models—such as the Dynamic Systems Theory (DST)—to effectively address its complex and varied manifestations.

Altogether, the discussion underscores that DST provides a flexible, evidence-based, and patient-centered model for managing rare and complex neurodevelopmental conditions. By focusing on task variability, sensory integration, visual-motor coordination, and multi-system interactions, DST-based physiotherapy interventions can produce measurable functional gains in motor control, balance, and independence. These findings advocate for a shift in paediatric physiotherapy from rigid, protocol-driven approaches toward dynamic, adaptable models that consider the whole child and their evolving interaction with the environment.

CONCLUSION

This case study illustrates the efficacy of the Dynamic Systems Theory (DST) framework in the physiotherapy management of a child with Joubert Syndrome (JS). DST enabled a multi-dimensional intervention addressing the child's neuromuscular, sensory, cognitive, and environmental challenges through task-specific, adaptable activities. The approach emphasizes the interaction of multiple subsystems in motor development, encouraging variability, adaptability, and self-organization. By addressing the unique motor, sensory, and cognitive challenges of Joubert Syndrome through DST-based interventions, physiotherapists can promote functional independence and improve the quality of life for affected individuals[13]. Additionally, the involvement of the family and adaptation of the child's environment are critical for achieving long-term success in therapy. As this case highlights, the application of DST in physiotherapy for JS can lead to significant motor improvements, demonstrating the theory's potential in treating complex motor disorders.

Future Applications

The findings of this study pave the way for integrating Dynamic Systems Theory (DST) into broader paediatric physiotherapy practice, especially for rare and complex genetic conditions like Joubert Syndrome. DST's adaptability allows for real-time adjustment to individual needs, making it suitable for use in tele-rehabilitation, community-based programs, and resource-limited settings. Future applications can include:

Development of DST-based physiotherapy protocols for children with multi-system involvement. Integration into early intervention services to improve developmental outcomes in at-risk infants.

Declaration by Authors

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