RESEARCH PAPER

Effect Of Pediatric Instrument-Assisted Soft Tissue Mobilization (Piastm) In A Child With Joubert Syndrome: A Case Report From Europe

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ABSTRACT

Background: Joubert syndrome is a rare genetic neurodevelopmental disorder characterized by hypotonia, oculomotor abnormalities, developmental delay, and impaired motor control. Children with Joubert syndrome often demonstrate significant postural instability, altered tone regulation and delayed motor milestones. Published evidence examining adjunctive physiotherapeutic interventions targeting fascial and proprioceptive systems in this population remains sparse.

Case Presentation: We report the case of a 2-year-5-month-old female diagnosed with Joubert syndrome due to a CPLANE1 gene mutation, presenting with global developmental delay, generalized hypotonia, joint stiff, oculomotor dysfunction, and impaired postural control. The child underwent Paediatric Instrument-Assisted Soft Tissue Mobilization (PIASTM) as part of a comprehensive rehabilitation approach, delivered over four consecutive days. Pre- and post-intervention visual and clinical observations were compared.

Results: Following PIASTM intervention, improvements were observed in ankle–foot alignment, heel contact during stance, symmetry of lower-limb loading, pelvic stability, trunk alignment, sitting balance and standing tolerance. These changes suggested improved tone regulation, enhanced proprioceptive input, and better motor organization.

Conclusion: This case highlights the potential role of PIASTM as an adjunctive physiotherapy intervention to improve postural alignment and functional motor control in children with Joubert syndrome. Further controlled studies are warranted to establish efficacy and long-term outcomes..

Keywords: Joubert syndrome; pediatric physiotherapy; PIASTM; hypotonia; postural control; neurodevelopmental disorders

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INTRODUCTION

Joubert syndrome (JS) is a rare autosomal recessive neurodevelopmental disorder classified among the ciliopathies, with an estimated prevalence of 1 in 80,000–100,000 live births. It is characterized by a distinctive midbrain–hindbrain malformation known as the molar tooth sign, resulting from hypoplasia of the cerebellar vermis and abnormal brainstem development. Clinically, children with JS commonly present with hypotonia, abnormal eye movements, impaired motor coordination, developmental delay, and challenges in postural control.

Hypotonia and truncal instability are hallmark features of JS and significantly interfere with the acquisition of gross motor milestones such as independent sitting, standing, and ambulation. The cerebellum plays a crucial role in motor planning, proprioceptive integration, and postural regulation; therefore, cerebellar dysfunction in JS results in impaired sensory–motor integration and fluctuating tone. These impairments frequently persist into childhood, highlighting the need for sustained, multidisciplinary rehabilitation strategies.

Physiotherapy management in Joubert syndrome primarily focuses on improving postural alignment, balance, proximal stability, and functional mobility through neurodevelopmental and task-oriented approaches. However, conventional therapy alone may be insufficient to address altered tone regulation, excessive joint mobility, and poor proprioceptive awareness commonly observed in hypotonic children. Enhancing afferent sensory input has therefore been recognized as a key therapeutic target in paediatric neurorehabilitation.

Paediatric Instrument-Assisted Soft Tissue Mobilization (PIASTM) is a paediatric-specific therapeutic approach designed to provide developmentally appropriate sensory and fascial input to support tone modulation, postural alignment, and motor organization in children with neuromotor and developmental disorders. PIASTM is a paediatric-specific therapeutic framework developed by Kanu Kaushik, designed to deliver developmentally

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appropriate sensory and fascial input in children with neuromotor disorders. Despite growing clinical use, evidence supporting PIASTM in rare neurodevelopmental conditions such as Joubert syndrome remains limited.

This case report aims to describe the observed effects of PIASTM on postural alignment, balance, and functional motor outcomes in a young child with Joubert syndrome, contributing preliminary clinical insight into its potential role as an adjunctive physiotherapy intervention.

CASE DESCRIPTION

Study Setting

The present case was assessed and treated in a PIASTM Therapy setting. All clinical evaluation and intervention procedures were conducted under the supervision of a trained PIASTM Practitioner.

Patient Information

Age at assessment: 2 years 5 months Sex: Female Month of assessment: November 2025

Primary Diagnoses

Joubert syndrome (CPLANE1 gene mutation) Intellectual developmental delay (ICD-10: F70)

Clinical Findings

Neurological examination performed by the treating physician revealed generalized hypotonia, more pronounced in the shoulder, trunk and pelvic area. A mild right-sided equinus tendency with minimal limitation of ankle dorsiflexion was observed. Deep tendon reflexes were symmetrical and brisk. The child was able to stand with support but demonstrated poor postural stability. Developmental quotient ranged between 40 and 50, consistent with moderate developmental delay.

Additional findings included oculomotor dysfunction with intermittent strabismus and hypermetropia, managed with occlusion therapy. Growth parameters were within age-appropriate percentiles. EEG findings were normal, with no history of seizures.

The child had previously participated in physiotherapy and electrostimulation programs and was enrolled in an integrated developmental kindergarten program including physiotherapy, special pedagogy, and vision-related support.

METHODS

PIASTM Intervention Framework

Paediatric Instrument-Assisted Soft Tissue Mobilization (PIASTM) was implemented as a specialized adjunct within the child's ongoing physiotherapy program. PIASTM represents a paediatric-specific, clinician-dependent neurorehabilitation framework requiring specialized training in PIASTM assessment and intervention.

The intervention was delivered exclusively by a paediatric physiotherapist formally trained in the PIASTM approach. Treatment decisions were guided by continuous clinical assessment of tone regulation, postural control, sensory responsiveness, and motor organization rather than predefined mechanical parameters. Specific technical details, including instrument handling characteristics, pressure modulation, stroke parameters, and sequencing, are training-dependent and therefore not disclosed.

PIASTM was administered across multiple structured sessions following an initial assessment. The intervention was delivered over **four consecutive days,** in accordance with the clinical PIASTM framework. Treatment was delivered using a **paediatric-specific instrument known as the Accel tool**, designed for controlled, developmentally appropriate sensory application in children. Each session had a **total duration of approximately 45 minutes,** during which the **entire body was addressed within a single treatment framework.** Session focus and therapeutic emphasis were dynamically adapted based on the child's tolerance, behavioural cues, and postural engagement. The intervention emphasized safety, comfort, and developmental appropriateness, with continuous monitoring for adverse responses. PIASTM was delivered as a standalone intervention focused on neuromuscular and sensory modulation, without concurrent integration of functional postural or task-oriented activities.

RESULTS

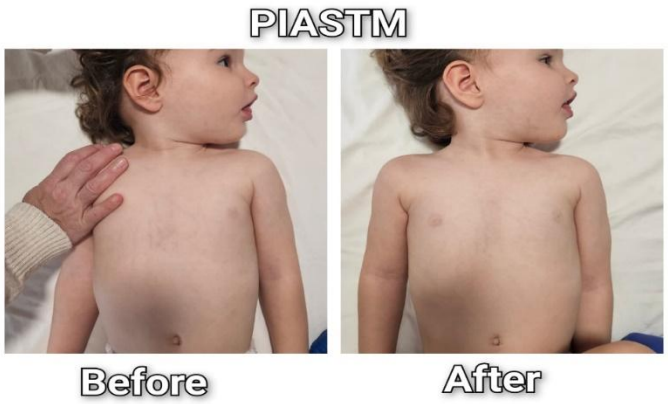
Following the implementation of PIASTM, clinically observable improvements were noted across multiple postural and functional domains. Outcomes were evaluated qualitatively using structured clinical observation and visual comparison of pre and post-intervention positioning, as standardized quantitative outcome measures were not formally employed.

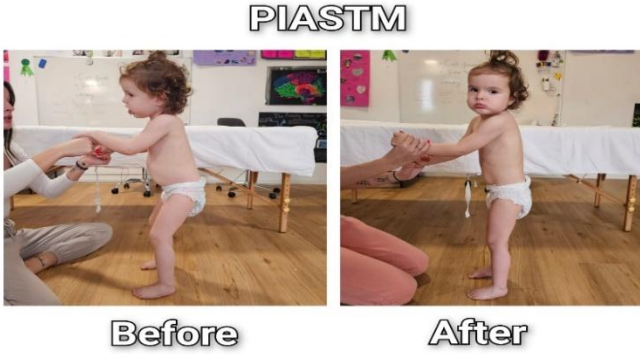
Improvements were observed in ankle dorsiflexion range, heel contact during stance, symmetry of lower-limb loading, pelvic alignment, trunk posture, sitting balance, standing tolerance, and transitional movements, suggesting improved tone regulation and postural organization.

Table 1. Physiotherapy Outcomes Following Pediatric Instrument-Assisted Soft Tissue Mobilization (PIASTM) in a Child with Joubert Syndrome

Outcome Domain	Pre-Intervention Findings	Post-Intervention Findings	Clinical Interpretation
Ankle dorsiflexion range	Mild limitation of right ankle dorsiflexion; tendency toward plantarflexion	Improved dorsiflexion range with more neutral ankle positioning	Suggests reduced distal tone and improved joint mobility
Heel contact in stance	Incomplete heel contact, unstable base of support	Consistent heel contact with improved weight acceptance	Indicates enhanced proprioceptive input and postural stability
Lower-limb alignment	Asymmetrical weight bearing,	Improved bilateral symmetry	Reflects better neuromuscul

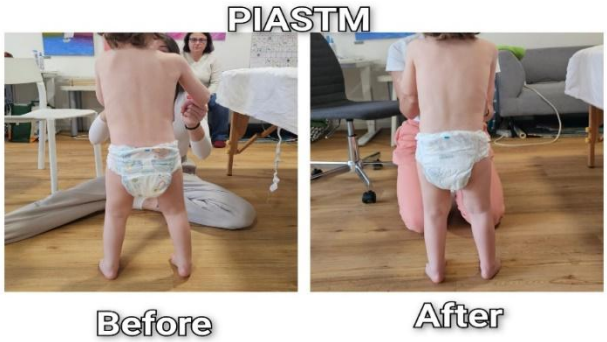
and symmetry	altered foot posture	during stance	ar coordination
Pelvic alignment and control	Pelvic instability with compensatory postures	More centered pelvis with reduced compensatory movements	Improved proximal stability
Trunk alignment	Rounded trunk posture, difficulty maintaining upright position	Improved upright trunk alignment	Suggests enhanced activation of deep postural muscles
Sitting balance	Reduced sitting balance, reliance on compensations	Improved sitting balance with better postural endurance	Improved sensory-motor integration
Standing tolerance	Limited standing duration, high reliance on external support	Increased standing tolerance with improved lower-limb extension	Indicates improved postural endurance
Transitional movements	Difficulty transitioning between positions	Improved control during transitions	Reflects improved motor planning
Overall tone regulation	Fluctuating tone, excessive joint flexibility	More organized movement patterns with improved tone regulation	Suggests improved central and peripheral integration






PIASTM
Before
After


PIASTM
Before
After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DISCUSSION

Children with Joubert syndrome exhibit complex motor impairments driven by cerebellar dysfunction, hypotonia, impaired proprioception, and delayed motor planning. In the present case, the child demonstrated classic features of JS, including generalized hypotonia, joint tightness or instability, and delayed standing ability.

Following the introduction of PIASTM, improvements were observed in distal alignment, proximal stability, and functional postural control. Improved heel contact and ankle positioning suggest enhanced sensory feedback from the ankle-foot complex, which is essential for postural stability in hypotonic children. Enhanced pelvic and trunk control likely reflect improved neuromuscular coordination and sensory-motor integration.

PIASTM is hypothesized to facilitate neuromuscular modulation through integrated stimulation of muscular and fascial tissues, enhancing afferent sensory input via cutaneous, myofascial, and proprioceptive mechanoreceptors, thereby supporting improved tone regulation and motor planning. Given the cerebellar involvement in Joubert syndrome, sensory-based adjunctive interventions may play a supportive role within a multimodal rehabilitation framework.

Although causality cannot be established from a single case, the observed changes are clinically meaningful and align with theoretical mechanisms proposed for sensory-modulating manual therapy techniques. PIASTM should be considered an adjunctive intervention requiring paediatric-specific training and advanced clinical reasoning.

CONCLUSION

This case report suggests that Pediatric Instrument-Assisted Soft Tissue Mobilization positively influence postural alignment, tone regulation and functional motor control in a child with Joubert syndrome. By enhancing proprioceptive input and supporting postural organization, PIASTM may represent a valuable adjunct to conventional pediatric neurorehabilitation in rare genetic neurodevelopmental disorders.

LIMITATIONS

This report is limited by its single-case design, which restricts generalizability. The intervention was delivered as part of a multimodal rehabilitation program; therefore, the specific contribution of PIASTM cannot be isolated. Outcome assessment relied primarily on qualitative clinical observation rather than standardized quantitative measures or blinded assessment. Additionally, longer-term follow-up and retention of observed changes could not be evaluated. Accordingly, these observations should be interpreted as preliminary and hypothesis-generating.

ETHICAL CONSIDERATIONS

Written informed consent was obtained from the child's parents or legal guardians prior to intervention and documentation. Consent included approval for the use of anonymized clinical data and images for academic and publication purposes. Patient confidentiality was maintained throughout. All procedures were conducted in accordance with the ethical principles of the Declaration of Helsinki. As this manuscript represents a descriptive single-case report without experimental manipulation, formal approval from an institutional ethics committee was not required.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

FUNDING

This research received no external funding.

DATA AVAILABILITY

All data generated or analyzed during this study are included in this published article.

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