

# Improvement in Balance and Functional Activity in Case Diagnosed with Juvenile Metachromatic Leukodystrophy: A Case Report

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**Abstract:** ***Purpose:** This case report investigated the effectiveness of the rehabilitation program in Balance & sit to stand from the floor independently in case with juvenile MLD. **Case description:** A 5-year-old boy participated in physical therapy, which included trunk stabilization, balance training and functional exercises for 12 weeks. **Assessments:** The pediatric balance scale was used as an outcome measure. **Result:** A positive change was observed in the pediatric balance scale, which was taken three times before, during, and after the protocol. **Conclusion:** Physical therapy improved in balance, lower limb strength, and most importantly able to achieve floor sitting to standing.*

**Keywords:** Juvenile MLD, Balance, Functional activity, sit to stand, Rehabilitation

## 1. Introduction

Metachromatic leukodystrophy (MLD) is a rare, autosomal recessive lysosomal storage disorder characterized by the accumulation of sulfatides due to arylsulfatase A deficiency, leading to progressive neurological symptoms.<sup>1,2</sup> It is estimated to affect 1.4 to 1.8 per 100,000 newborns.<sup>3</sup> The disorder manifests in three clinical forms: late infantile, juvenile, and adult-onset, with juvenile MLD typically presenting between ages 3 to 16 years.<sup>4</sup>

Current treatment options for MLD remain limited. Hematopoietic stem cell transplantation is employed to slow disease progression; however, its efficacy is still debated due to potential risks and varying long-term outcomes.<sup>5</sup> Emerging therapies, such as enzyme replacement therapy and gene therapy, show promise but require further investigation.<sup>6</sup>

The impact of physical therapy on improving functional outcomes in children with neurodegenerative disorders, including MLD, has gained attention. Structured rehabilitation can enhance balance and mobility, addressing significant motor deficits experienced by affected children.<sup>7,8</sup>

MLD has three types: 1. Late infantile onset = 6 months - 2 years; 2. Juvenile onset = 3 years - 16 years; and 3. Adult onset = > 16 years.<sup>9</sup>

In this article, we present the clinical findings of Children with juvenile MLD and report the 12-week physical therapy program's effectiveness in improving balance and sit-to-stand function. Informed consent was obtained from the children's parents.

## 2. Case Report

### History

This case report followed a 5-year-old male child SS who was born by full-term normal delivery and had a spontaneous cry and had no complications during delivery the patient was growing normally by achieving all normal milestones was able to walk run play with friends and talk with friends and family like a normal child.

On 24/June/2024 in the afternoon around 1 pm patient was playing with friends on a rainy day when he fell in the mud his mother brought him home where the patient got fever and fever was increasing severely so her mom took him to a local hospital where he was getting seizures 2 times which last 5 minutes so doctor refer patient to Sparsh hospital where he was admitted in ICU and blood test was done 28/June/2024 on 4:41 pm (leucocyte and neutrophils was increased, MCV reduced). On 30/June/2024 blood test was done at 8:48 am (only MCV reduced) and shifted to the ward, on 02/July/2024 MRI of the brain was done which showed symmetrical frontoparietal periventricular white matter lesions with predominant cortical atrophy MRI suggested leukodystrophy and the differential diagnosis was metachromatic, the patient was referred to KEM hospital, Mumbai where they got admitted on 10/July/2024 and on 11/July/2024 patient blood was send for genetic studies in center of DNA, fingerprint and diagnostics, Hyderabad where the date of collection was 28/July/2024 and reported on date 23/August/2023 that ASA enzyme was 1.17 mmol/hr/mg (normal range = 25 - 80 mmol/hr/mg) ASB enzymes was 71.45 mmol/hr/mg (normal range = 115 - 226 mmol/hr/mg).

From 18/July/2024 patient started physiotherapy treatment in Neuro OPD at MIMSR Medical College, Latur. Where patient came with chief complaints of walking with support with a wide base of gait pattern, not being able to stand more than 10

seconds independently without support, difficulty maintaining balance (frequent falls while walking), and not being able to sit to stand from floor and chair independently without use of support and show mental impairment of not speaking independently and being quit all-time at home and neuro OPD and only use to repeat words which therapist or mother use to say patient was not interacting as patient use to do before like normal child.

### 3. Assessment

1)The patient is oriented to time and person but not to place and takes a longer time to understand and respond, 2)The memory of the patient is poor patient only repeats words that are said by the therapist or mother every time even if a patient is told to not repeat words patient say a word which therapist or mother say, 3) all 12 cranial nerve was intact, 4) deep tendon reflex was 1+ both lower limb and upper limb and Babinski reflex was negative, 5) superficial sensation was intact in both upper limb and lower limb, deep sensation pressure and vibration was intact, posture and movement sensation and cortical sensation patient didn't respond, 6) muscle tone is normal, 7) all active range of motion intact in both upper limb and lower limb, 8) manual muscle testing is grade 3 in both lower limb and grade 4 in both upper limbs, 9) Pediatric balance scale = 10/56(0 to 20 high fall risk).

### 4. Procedure

Ethical approval from the Institutional Ethical Committee was not needed and the patient's mother gave an informed written consent. His demographic data, physical examination, paediatric balance scale was recorded. from 18 July to 31<sup>st</sup> July 2024 physiotherapy treatment was VMO exercises and dynamic quadriceps exercise by kicking ball 10 reps 2 set, wall bar hand support squatting with mild assistance by therapist to strengthen gluteus maximus and quadriceps muscles 10 reps 2 set, clamp shell exercises to strengthen gluteus Medius muscle with use of yellow TheraBand 10 reps 2 sets, prone knee bend with use of yellow TheraBand by asking touch heel on my hand above his gluteus area 10 reps 2 set to strengthen hamstring muscles, heel raise exercise in standing with support by holding both hand 10 reps 2 sets, wobble board exercise with hand support holding wall bar and trying to maintain balance giving mild support from pelvis posteriorly to stand 15 reps side to side movement on wobble board to strengthen ankle muscle and improve ankle stability.

From 1<sup>st</sup> August to 21<sup>st</sup> August 2024 instead of yellow TheraBand, we used half kg weight cuff while doing VMO and dynamic quadriceps exercises, clamp shell exercises and prone knee bend exercises (all exercise 10 reps 2 sets) to improve strength from grade 3+ to grade 4 of manual muscle testing, new treatment was added was:- a) side way walking (strengthen gluteus Medius muscle) with use of yellow TheraBand tied on both leg above ankle with support by both hand of therapist walking together with patient sideways from start point to end point completing at 20 steps continuing up to completion of 2 laps b) bridging exercises 10 reps 2 sets (strengthen gluteus maximus), c) sit and reach for circle and pick from floor (strengthen tibialis anterior and quadriceps in reaching activity) and stand with support of one hand and reaching overhead for cone by standing on toe (strengthen

quadriceps and soleus muscles) to put circle on cone and sit back slowly with use of one hand support dosage 10 reps 2 sets, d) step up and step down exercise (strengthen quadriceps and gluteus maximus muscle) with use of wooden box height of patient knee level 10 reps with each leg 2 sets, e) wobble board exercise with support of both hand anterior and posterior movement of wobble board (strengthen and improve stability in dorsiflexor and plantar flexor muscle) 15 reps, f) walking on straight line on TheraBand by placing heel in front of opposite leg toe like tandem walking with support of one hand (to improve balance and reduce wide base of gait pattern) 20 steps 2 laps.

Patient progression at end of 21<sup>st</sup> august 2024 was patient was able to sit to stand from chair independently, able to walk with wide base of gait independently without support but use to lose balance while changing direction or turning, able to stand independently more than 30second without support, able to transfer but use to use hand while sitting, able to pick object from floor while standing, MMT was grade 4 in both lower limb, paediatric balance scale = 33/56(medium fall risk).



Figure 1: Sit to stand from chair

Limitation patient was not able to stand from floor and use to crawl on floor to play

From 21<sup>st</sup> August to 12<sup>th</sup> September 2024 physiotherapy treatment was:- a) wall bar climbing up and coming down with therapist support on pelvis 5 reps 2 set (strengthen gluteus muscle, quadricep muscle and gastrocnemius and soleus muscle), b) stair climbing and descend down from stairs 10 reps without support, stepping up and down from low height block below knee level 10 reps 1 set independently with minimal support by 1 hand, c) training to pick circle from start of TheraBand and do tandem walking without support and put a circle on cone on the end of TheraBand 5 reps 2 sets (improve balance and improve trunk extension), d) half squat on balance stabilizer 10 reps with 1 hand support (improve

balance), e) trunk extension and abdominal curls and sitting side to side roll on vestibular ball 10 reps each exercise 1 set (strengthen trunk extensor and abdominal muscle and improve stability and balance).

Progression [patient was able to sit to stand from floor independently and use to stand for playing] paediatric balance scale= 41/56(low fall risk).



**Figure 2:** Floor sitting to standing

## 5. Discussion

This case report presents a 5-year-old boy diagnosed with juvenile Metachromatic Leukodystrophy (MLD), who underwent a structured physical therapy program over 12 weeks, focusing on improving balance and the ability to transition from sitting on the floor to standing independently. The results indicated significant positive changes in balance and functional mobility, as quantified by the Pediatric Balance Scale (PBS).

### Overview of Findings

The case described reflects a comprehensive physical therapy approach tailored to the unique challenges posed by juvenile MLD. The progressive rehabilitation protocol included exercises focused on muscle strengthening, balance improvement, and functional mobility. Notably, the initial assessment showed the child had a PBS score of 10/56, indicating a high risk of falls, and post-intervention, this score improved significantly to 41/56, suggesting a low fall risk. This outcome emphasizes the potential effectiveness of targeted physical therapy in managing the motor deficits associated with MLD.

### Step-by-Step Training Approach

The structured progression of exercises tailored to the child's needs was a key feature of the treatment. Initially, exercises

were centered around fundamental stability and muscle strengthening, including tasks like squatting and dynamic quadriceps exercises that targeted the major muscle groups necessary for standing and balance. As the child's abilities progressed, more complex activities were introduced, such as sidestepping with band resistance and tandem walking, which require greater coordination and dynamic balance.

These adaptations in the physiotherapy regimen not only improved muscle strength—from manual muscle testing of grade 3 to grade 4 in lower limbs—but also enhanced the child's functional abilities, enabling him to independently sit to stand from a chair and the floor, tasks which are critical for his mobility and participation in daily activities.

## 6. Limitations of the Study

While the positive outcomes from this targeted rehabilitation program are encouraging, it is essential to acknowledge the limitations inherent in this case report. The findings stem from a single patient experience, limiting the generalizability of the results. Additionally, the absence of a control group makes it difficult to ascertain the effectiveness of the intervention in a comparative context.

Another limitation is the short duration of follow-up; prolonged assessments could provide better insights into the sustainability of these gains. Further investigations with larger sample sizes and control conditions are warranted to establish the efficacy of such rehabilitation protocols on a broader scale.

## 7. Conclusion

In conclusion, the case of this 5-year-old Child illustrates that a carefully structured rehabilitation program can lead to significant functional gains in balance and mobility for patients with juvenile MLD. Despite the challenges presented by this rare and complicated disorder, targeted physical therapy can foster independence and improve motor skills crucial for everyday life. Continued research in this domain may provide deeper insights into effective management strategies for similar cases in the future.

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