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Case Report

Functional recovery in a rare case of an adult patient with leucodystrophy

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ABSTRACT

The adulthood leukodystrophy is an uncommon clinical condition. Physiotherapy treatment provided to this referred patient resulted in a significant functional recovery within a short span of time inspite of gross lesion. A 34-year-old male patient diagnosed as a case of leucodystrophy was referred for Physiotherapy after 2 years of initial diagnosis physiotherapy progress consisted of increasing muscle strength, reducing tightness and improving postural stability; also to improve mobility for 5 months. After taking the pre and post assessment values of MAS, FIST, mFRT and FIM, improvement through the post assessment values showed significant functional recovery within 5 months. Significant functional recovery after 2 years of diagnosis of leucodystrophy provides promising scope of physiotherapy in future cases of leucodystrophy.

Keywords: Leucodystrophy, Rehabilitation, Physiotherapy, Functional recovery

INTRODUCTION

According to current definitions, leukodystrophies are described as genetically based illnesses that largely impact the central nervous system's white matter, regardless of the specific molecular process, patient age group, or disease course involved. While early infancy and childhood are the main times when these illnesses emerge, they can also affect adults, who can sometimes present with different clinical and imaging findings from children. Leukodystrophies are becoming more widely recognized for their potential to first appear in adulthood. Adult leukodystrophies usually are progressive diseases, presenting with movement disturbance, vision problems, hearing impairment, imbalance, memory loss, behavioural changes, and attention deficits.[1]

Adult leukodystrophies are currently classified into about 20 different syndromes; these conditions differ from other inherited leukoencephalopathies in that they entail additional involvement of grey matter structures or non-cerebral organs. Furthermore, phenotypes in adulthood leukodystrophies are incompletely characterized in approximately 50% of all cases, resulting in a high estimated number of unreported cases and prolonged diagnostic procedures.[2] Although very few cases with adult leukodystrophy have been reported in literature. Its functional recovery over a period of time and role of physiotherapy is not mentioned.

A case report is described here of an adult patient with leucodystrophy referred for physiotherapy treatment. Functional status at baseline and after 5 months of treatment is reported here. Although full recovery could not be achieved but patient was independent in most of the daily activities at the end of 5 months.

CASE REPORT

A 34-year-old male was brought to the physiotherapy outpatient department (OPD) with the complaints of difficulty in bed mobility, difficulty in independent sitting, difficulty in lower limb dressing and inability to walk, difficulty in holding objects for prolonged period with the

left hand. The patient was apparently alright 3 years back when he had a fall from a tree about 20 feet in height. The patient was in a coma for about a month. When he regained consciousness he was able to move his bilateral upper limb and lower limb. The patient could walk using a walker/cane then for about six months. However, he began experiencing progressive lower limb weakness (distal> proximal) and also in the left upper extremity. The patient was bed ridden for the past 1.5 years. And was brought to the OPD from the suggestion of a relative for physiotherapy rehab.

He was a farmer for 10 years and is currently unable to resume his job. On examination patient's motor examination revealed, presence of grade 1 spasticity on MAS in left elbow flexor. According to MRC grading the muscle strength is mentioned in Table 1. Strength of abdominal muscles was fair and back extensor muscles were poor. The reflex testing elicited with positive Babinski sign for bilateral plantar response, normal upper limb deep tendon reflexes bilaterally, and hyperreflexia seen for lower limbs.

Table 1: Timeline of progression of disease and the functional capacity from onset till referral for physiotherapy program.

Dates	Events
24 April 2020	Fall from 20 feet tree
April 20'to May 20'	Comatose- 1 month
June 2020 to August 2020	Ambulation with walker for next 3 months
September 2020 to November 2020	Ambulation with stick for next 3 months
From December 2020	Progressive weakness observed
Since January 2021	Reduced ADLs and ambulation to minimum

Non equilibrium coordination testing revealed that movements were smooth and controlled, accurate for bilateral upper limb and they were not smooth and accurate for bilateral lower extremities whereas Equilibrium tests could not be assessed as patient has difficulty in standing. He had tightness of bilateral hamstring, tendoachilles and both side hip adductor muscle. Postural examination revealed forward head posture with left-sided thoracolumbar scoliosis along with right sided pelvic hike and congenital talipes cavovarus. For hand functions, he preferred radial grasp with the left hand to hold larger objects and grip was fair unilaterally (left) and the right hand functions were normal.

Functionally patient was able to sit with support and stand with maximum difficulty, he was able to perform activities of dressing for upper extremity and trunk but had difficulty with donning and doffing for lower extremity, for toileting activities wheelchair ambulation was required, and had independent eating and drinking activity.

Figure 1: Bilateral restriction diffusion with T2 and FLAIR hyperintensities without blooming on SWI seen at bilateral centrum semiovale, along the corticospinal tract, posterior limb of internal capsule, ventrolateral aspect of midbrain and pons.

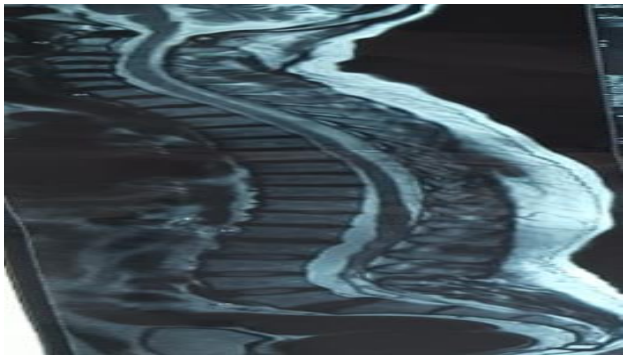

Figure 2: Posterocentral disc bulge at C3-4 and C4-5 levels, indenting anterior subarachnoid space. Lumbosacral spine shows changes of disc dessication with posterior-central disc bulge at L5-S1 level, indenting thecal sac and traversing nerve roots.

Physiotherapy intervention

The patient was referred to physiotherapy for rehabilitation to improve activities of daily living in February 2023. His lower limb rehabilitation progressed well with range and strength returning gradually. A significant proportion of his physiotherapy therefore focused on increasing muscle strength, reducing spasticity and tightness along with improving postural stability.

Goals

The primary goal was to improve lower limb mobility with minimal assistance as he had difficulty to actively move the lower limbs. Next was to reduce the tightness of hamstring muscles of both the limbs. Improving posture and emphasis on developing erect posture was one of the goals to be targeted. Further goal was to increase the muscle strength of the left hand muscles, hip and knee musculature along with the trunk muscle strength. Transfer training for sit to stand and bed mobility was also the focus.

1st month - 3rd month

In this early stage we established a baseline measure of the patient's neurology in terms of strength, functional range, posture and multidirectional stability along with balance in sitting and standing position. Supine to side lying, Supine to prone, Side to sit activities which were re-educated with the help of: rolling, pelvic PNF, supine to sit, and side weight bearing on elbows. For difficulty in moving lower limbs the treatment was performing and guiding Active assisted ROM exercises to improve the movement performance. For the tightness of the hamstring and trunk muscles due to left-sided thoracolumbar functional scoliosis, stretching- for 30 seconds per repetition for 5 times and application of hot packs 5 mins for each limb. Positioning with the help of pillows was advised. Visual and verbal feedback was reinforced. To increase the strength of hand muscles resistive strength training programme with manual resistance; also for the hip and knee muscles strength training 1st- started with static holds of 10 sec. To strengthen the trunk muscles, exercises given were- abdominal curls, oblique curls, and prone on elbows. The normalization of tone of elbow flexors was treated with: resistive D1 extension pattern of PNF, and eccentric strength training for elbow flexors. Transfers were trained with: sit to stand training in parallel bars with mirror for feedback, multiple angle holds for sitting, and supported standing including functional movements such as squats and bridging.

4th month - 5th month

The patient's lower limb rehabilitation at this point was centered on introducing increasingly difficult functional exercises to test strength and balance, like sit-to-stand and assisted walking, along with a steady increase in resistance. Range of motion improvement was progressed to using skates for lower limbs. For the tightness of the lower limb muscles and the trunk muscles the stretching programme was continued as self-exercise program with advise for postural correction. Hand intrinsic muscle strengthening was done with the use of finger and hand grip exerciser. The strengthening of the knee and hip musculature as well as trunk muscles was proceeded with increasing the subjective manual resistance; and increasing the hold time for increasing the trunk muscles strength. Also, for improvement of the transfer of sit to stand which he could perform with mild difficulty was done with feedback and increased repetitions for multiple angle holds and increased supported standing time. His rehabilitation is still ongoing where we are monitoring where his lower limb rehab will continue to be progressed by increasing his strength, balance and control.

Outcome measures

We used FIST to assess seated postural control which is a measure of functional sitting balance is reliable, valid, and easy to administer in patients with stroke.[3] The modified functional reach test (MFRT) was used as the objective measure for dynamic sitting balance and appears to provide reliable measurements of sitting balance in nonstanding persons with SCI.[4]

FIM was used to reliably document and report a patient's level of disability.[5] To assess motor function Motor assessment scale was used it is a reliable and validated test of motor function in stroke patients.[6]

Table 2: Manual muscle strength grade of muscles of upper and lower extremities at baseline and after 5 months of treatment.

Muscle group UL	Right	Left	Muscle group LL	Right	Left
Shoulder			**Hip**		
Flexors	4+/5	4/5	Flexors	3-/5	3-/5
Extensors	4/5	4/5	Extensors	3-/5	3-/5
Abductors	4/5	4/5	Abductors	2/5	2/5
Adductors	4/5	4/5	Adductors	2/5	2/5
Elbow			**Knee**		
Flexors	4/5	4/5	Flexors	3-/5	3-/5
Extensors	4/5	4/5	Extensors	3/5	3/5
Wrist			**Ankle**		
Flexors	5/5	4/5	Plantarflexors	1/5	1/5
Extensors	4/5	4+/5	Dorsiflexors	2/5	2/5
Intrinsics			**Toes**		
Finger flexors	5/5	4/5	Flexors	5/5	3/5
Finger extensors	5/5	4/5	Extensors	3/5	2/5
Thumb muscles	Normal	Fair	-	-	-
Abductor digiti minimi	5/5	3/5	-	-	-
Dorsal and palmar interossei	5/5	3/5	-	-	-
Lumbricales	5/5	2/5	-	-	-

Table 3: Improvement in functional capacity using objective outcome measures, over a period of time.

Component	Measurement method	March 2023	April 2023	May 2023	June 2023	July 2023	Change from March to July (%)
Sitting balance	Functions in sitting test (FIST) test	38/56	42/56	52/56	52/56	55/56	30.35
	Modified functional reach test (mFRT) (cm)						
	Forward	25	29	31	35	37	25.5
	Right	12	18	22	24	25	46.4
	Left	12	16	19	18	22	34.4
Functional independence measure (FIM)	**FIM assessment tool**						
	Self-care	23/49	25/49	26/49	27/49	28/49	10.2
	Spincter control	14/14	-	-	-	-	Not affected
	transfers	6/21	8/21	11/21	15/21	18/21	28.5
	locomotion	5/14	6/14	7/14	8/14	9/14	28.5
	Cognitive tasks	14/14	-	-	-	-	Not affected
	Social cognition	21/21	-	-	-	-	Not affected
Motor assessment scale (MAS)	**MAS scale**						
	Supine to side lying	4/6	4/6	5/6	5/6	5/6	16.7
	Supine to sit	3/6	3/6	4/6	5/6	5/6	16.66
	Balanced sitting	3/6	3/6	4/6	4/6	5/6	33.34
	Sitting to standing	2/6	2/6	2/6	3/6	3/6	16.7
	Walking	0/6	0/6	0/6	2/6	3/6	50
	Upper arm function	2/6	2/6	4/6	5/6	5/6	50
	Hand movement	6/6	-	-	-	-	Not affected
	Advanced hand activities	6/6	-	-	-	-	Not affected
	General tonus	6/6	-	-	-	-	Not affected

DISCUSSION

In this case reported, we have reported the clinical symptoms of a 34-year-old male with leukodystrophy and described the effect of physical therapy and rehabilitation program, which was continued 7 days per week for month. After the treatment, positive improvements were noted both in body structures and functions and in the level of activity and participation. When the case presented to our clinic, he could perform bed mobility with maximum difficulty and perform ADL's with maximum assistance. After 2 months of physical therapy, he could perform bed mobility without assistance and only needed mild assistance for his ADL's. He could sit for longer durations and dynamic sitting balance was improved.

Presentation of leukodystrophies may occur at any age. There is inverse correlation of age of onset with the disease severity and rate of progression. Leukodystrophies have been found to be progressive in most of the cases. The prognoses of this disease are associated with episodic deterioration which makes prediction difficult may not always be poor. The course of the disease is typically characterized by episodes of neurological decline, provoked by external or internal stressors, followed by partial recovery. Patients show highly variable disease signs with or without any disability for decades.

This is the first case report investigating the effect of the physical therapy and rehabilitation program on body structures and functions and the level of activities and participation in an adult with leukodystrophy. Our case had positive improvements in muscle strength, muscle tone, trunk control (TCMS), and balance after an 8-week physical therapy program.

Dynamic stretching reduced the tightness in B/L lower limb hamstrings and adductors which was also found effective in a study by Iwata et al who found that hat dynamic sitting caused a sustained reduction in passive stiffness of the hamstrings and increase in knee ROM and Hammer et al who reported a significantly improved ROM post 3 different types of stretches.[7,8]

We observed improvement in his sit to stand actively after training which was similar to the findings of Hyun et al who in their study found that the real time visual biofeedback sitting to stand training significantly improved the lower extremity muscle strength, balance, gait, and quality of life of the participants compared to that observed with the sit-to-stand training.[9]

Scoliosis correction was obtained with interventions such as self-correction through active muscle contraction and self-elongation of the torso muscles in a corrected posture which was also found effective in a systematic review by Bettany-Saltikov et al.[10]

Reduction in left elbow flexor spasticity was obtained, eccentric strengthening was provided along with resisted PNF for the same, the results are consistent with the findings of Manca et al who in a study found that eccentric

contractions proved feasible and safe in the management of spasticity of the elbow flexors muscles in people with multiple sclerosis.[11]

The essential goal was to enable the patient to achieve the greatest possible independence by improving the performance and execution of their motor skills.[12]

CONCLUSION

This study concluded that vigorous problem-specific physiotherapy intervention resulted in major functional improvement in this adult patient of leucodystrophy.

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REFERENCES

1. Resende LL, de Paiva ARB, Kok F, da Costa Leite C, Lucato LT. Adult Leukodystrophies: A Step-by-Step Diagnostic Approach. Radiogr Rev Publ Radiol Soc N Am Inc. 2019;39(1):153-68.
2. Köhler W, Curiel J, Vanderver A. Adulthood leukodystrophies. Nat Rev Neurol. 2018;14(2):94-105.
3. Gorman SL, Radtka S, Melnick ME, Abrams GM, Byl NN. Development and validation of the Function In Sitting Test in adults with acute stroke. J Neurol Phys Ther JNPT. 2010;34(3):150-60.
4. Lynch SM, Leahy P, Barker SP. Reliability of measurements obtained with a modified functional reach test in subjects with spinal cord injury. Phys Ther. 1998;78(2):128-33.
5. Keith RA, Granger CV, Hamilton BB, Sherwin FS. The functional independence measure: a new tool for rehabilitation. Adv Clin Rehabil. 1987;1:6-18.
6. Jl P, Sl W. Motor assessment scale for stroke patients: concurrent validity and interrater reliability. Arch Phys Med Rehabil. 1988;69(3 Pt 1).
7. Iwata M, Yamamoto A, Matsuo S, Hatano G, Miyazaki M, Fukaya T, et al. Dynamic Stretching Has Sustained Effects on Range of Motion and Passive Stiffness of the Hamstring Muscles. J Sports Sci Med. 2019;18(1):13-20.
8. Hammer AM, Hammer RL, Lomond KV, O'Connor P. Acute changes of hip joint range of motion using selected clinical stretching procedures: A randomized crossover study. Musculoskelet Sci Pract. 2017;32:70-7.
9. Hyun SJ, Lee J, Lee BH. The Effects of Sit-to-Stand Training Combined with Real-Time Visual Feedback on Strength, Balance, Gait Ability, and Quality of Life in Patients with Stroke: A Randomized Controlled Trial. Int J Environ Res Public Health. 2021;18(22):12229.
10. Bettany-Saltikov J, Parent E, Romano M, Villagrasa M, Negrini S. Physiotherapeutic scoliosis-specific exercises for adolescents with idiopathic scoliosis. Eur J Phys Rehabil Med. 2014;50(1):111-21.
11. Manca A, Martinez G, Aiello E, Ventura L, Deriu F. Effect of Eccentric Strength Training on Elbow Flexor Spasticity and Muscle Weakness in People With Multiple Sclerosis: Proof-of-Concept Single-System Case Series. Phys Ther. 2020;100(7):1142-52.
12. Lakshmanan R, Rajalaxmi V. A Comprehensive Physical Therapy Approach in Late Infantile Form of Metachromatic Leukodystrophy – A Case Study. Res J Pharm Technol. 2020;13(5):2214-6.
13. Harishchandre M, Mahajan S, Ganvir S, Tungikar. Influence of Myofacial Release and Stretching to Relieve Spasticity in Patients with Stroke – A Systematic Review. Int J Innov Sci Res Technol. 2022;7(11):1664-8.

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