International Journal for Multidisciplinary Research (IJFMR)
E-ISSN: 2582-2160 ● Website: www.ijfmr.com ● Email: editor@ijfmr.com

Effectiveness of Customised Physiotherapy in Managing Motor Neuron Disease Variants: A Case Series

Ms. Kanna Muni Ramya[1], Dr. Vadlamudi Srikumari[2], Dr. Gaali Kameshwari[3], Dr. Koneti Madhavi[4]

[1]MPT 2nd Year Neurology, Physiotherapy, COP, SVIMS
[2]MPT (Neuro), Ph.D, Associate Professor, Physiotherapy, COP, SVIMS
[3]MPT (Neuro), Assistant Professor, Physiotherapy, COP, SVIMS
[4]MPT (Ct),Ph.D, Principal, Physiotherapy, COP, SVIMS

Abstract:
Motor neuron disease (MND) encompasses a group of chronic, sporadic, and hereditary neurological disorders marked by progressive motor neuron degeneration, affecting upper, lower, or both types of motor neurons. While diagnosis is often clinically evident, it plays a critical role in formulating long-term physiotherapy plans for ongoing management. This case series presents three patients with atypical MND variants, initially suggestive of other conditions, resulting in delayed diagnosis. Once confirmed,customised physiotherapy interventions were implemented to address respiratory compromise, generalized limb weakness, and speech/swallowing difficulties.Regular assessments allowed for ongoing adjustments to the treatment approach, focusing on a comprehensive strategy aimed at improving the patient's overall well-being. Outcome measures and follow-up evaluations showed improvements in functional scales and manual muscle testing. This case highlights the importance of customised physiotherapy in achieving optimal functional outcomes and enhancing the quality of life for patients facing the complex challenges of bulbar MND.(1)A 75-year-old male patient diagnosed with bulbar motor neuron disease (MND) - ALS presented with worsening respiratory issues, generalized limb weakness, and challenges with speech and swallowing. A customised physiotherapy plan was developed to enhance his quality of life. (2)A 62-year-old male patient diagnosed with bulbar motor neuron disease (MND) - ALS presented with mild respiratory issues, generalized asymmetrical limb weakness, and difficulties with speech. A customised physiotherapy was planned. (3)A 13-year-old male patient diagnosed with SMA Type III presented with severe respiratory difficulties, including pectusexcavatum, generalized muscle atrophy of the limbs and trunk, widespread fasciculations, tongue fibrillations, and speech challenges. A customised physiotherapy plan was created to improve his quality of life and support long-term well-being.

Introduction:

Motor neuron diseases (MNDs) are a group of progressive neurological disorders that destroy motor neurons, the cells that control skeletal muscle activity such as walking, breathing, speaking, and swallowing. This group includes diseases such as amyotrophic lateral sclerosis, progressive bulbar palsy, primary lateral sclerosis, progressive muscular atrophy, spinal muscular atrophy, Kennedy's disease, and post-polio syndrome.

Amyotrophic lateral sclerosis (ALS), formerly known as Lou Gehrig's disease, can affect the upper and/or lower motor neurons. It causes rapid loss of muscle control and eventual paralysis. **Spinal muscular atrophy (SMA)** refers to a group of hereditary diseases that affect lower motor neurons. The most common form is caused by a mutated or missing gene known as the survival motor neuron gene 1 (SMN1), which causes the neurons to deteriorate, producing muscle weakness and wasting.SMA may present at birth or in infancy, in adolescence or in adulthood, tending to be most severe in infantile onset cases.

The Global Burden of Disease project recently estimated the MND all-age global prevalence to be 4-5 per 100,000 persons (1).The incidence of SMA is one in 10,000 live-born babies and is the most common cause of death in the infantile age group. In the West, the carrier frequency of the SMA is 1 in 50. In a recent Indian study, however, the SMA carrier frequency was 1 in 38 (2). About 20% of patients have MND with bulbar onset.

MND typically presents with slurred speech from impaired tongue movement, progressing to tongue wasting, fasciculations, and later dysphagia. Classification is based on onset site (spinal or bulbar), diagnostic criteria adherence (El Escorial/Airlie House), and heritability pattern (sporadic vs. familial)(3). Sporadic ALS commonly begins between ages 58–63, while familial ALS presents between 40–60 years (4).

Motor neuron diseases (MNDs) exhibit varied and uncommon symptoms, necessitating customised physiotherapy to address individual needs such as respiratory issues, limb weakness, and speech or swallowing difficulties. This case series emphasised the vital role of customised physiotherapy in enhancing functional recovery and improving quality of life in individuals with MND.

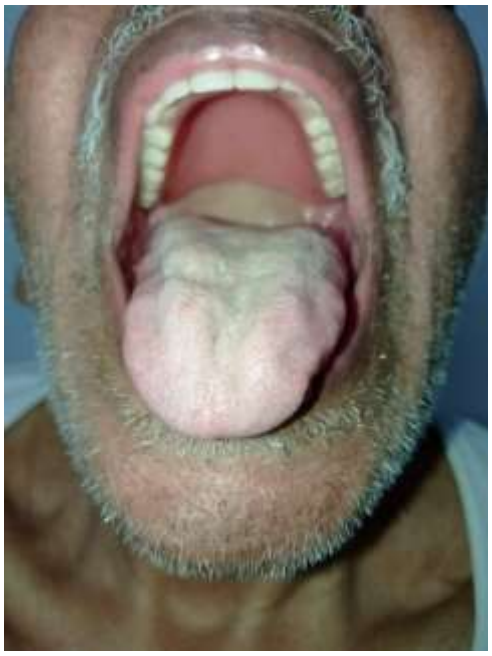
Case presentation:

Case-1

Patient information

This is the case of a 75-year-old male patient, a retired army officer, right-hand dominant, residing in Hyderabad. The information was provided by the patient. He had a 20-year history of hypertension and was on regular medication. Since 2022, he gradually experienced slippage of footwear while walking, followed by progressive difficulty in rising from a sitting position, climbing stairs, walking, holding objects, and feeding himself.

With these complaints, he visited KIMS Hospital. After undergoing an MRI of the spine, he was diagnosed with Prolapsed Intervertebral Disc (PIVD) at L3-L4 and L4-L5 levels with nerve root compression, and surgery was advised. He underwent spinal fixation surgery at the L3-L4 and L4-L5 levels in February 2024.Post-surgery, the patient remained stable for about three months. However, he later developed progressive weakness in both upper and lower limbs, along with slurred speech. He returned to KIMS Hospital, where Electromyography shows s/o neurogenic process involved with active and chronic reinnervation changes in right tibialis anterior & Extensor digitorum communis. All other muscles showed only chronic reinnervation changes. Diagnosis:by revised EI Escorial criteria: Definite ALS and was advised to continue physiotherapy treatment at home.Following the suggestion of relatives, the patient later visited SV Ayurvedic Hospital in January 2025.



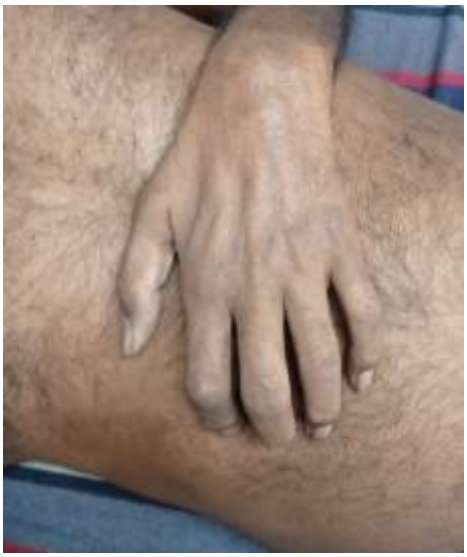
Picture 1: Tongue Fasciculations

Case-2

Patient information

This is a 62-year-old male patient, a retired assistant manager, right-handed, residing in Pulivendula. The information was provided by the patient himself. He has a medical history of Type 2 Diabetes Mellitus for the past 20 years and is on regular medication for the condition.

In 2022, the patient developed ankle pain and initially received medication at a local hospital, but the symptoms persisted. A neurologist later prescribed additional medications and advised three-month follow-ups. Over time, the patient experienced progressive weakness in all limbs, more on the left side, along with fasciculations in the trunk and limbs. This led to worsening symptoms and muscle wasting in the left upper and lower limbs.The patient visited CITI Hospital in Hyderabad, where investigations, including EMG and MRI, were conducted. Electroneuromyography revealed a neurogenic lesion involving the left C5–C8 and L4–L5 segments, with signs of acute and chronic denervation and reinnervation. MRI of the spine showed mild degenerative changes in the lumbar region, disc bulges at L1–L2, L2–L3, L3–L4, and L4–L5 with facetalarthropathy, and mild disc bulges at C3–C4, C4–C5, C5–C6, and C6–C7, indenting the thecal sac and spinal cord. Brain MRI showed diffuse age-related cerebral atrophy, old lacunae, and small vessel ischemic changes in the right external capsule. The patient was diagnosed with Motor Neuron Disease–ALS and prescribed Riluzole along with home-based physiotherapy. On January 30, 2025, following relatives' advice, he was taken to SV Ayurvedic Hospital.



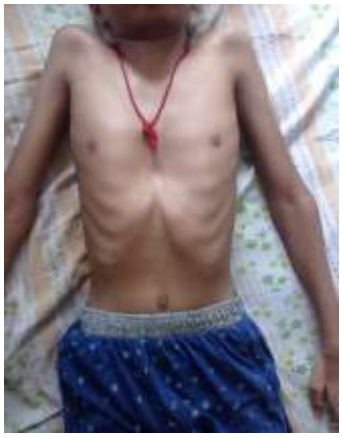
Picture 2: Wasting of intrinsic muscles of hand

Case-3

Patient information

This is a 13-year-old male child, a student, right-handed, residing in Tirupati. The information was provided by the mother. The child was normal until 2014, when at the age of 4, he developed a fever accompanied by seizures. He was admitted to a local private hospital and received treatment. However, weakness in both the upper and lower limbs persisted.

In 2017, the child was taken to CMC Vellore Hospital, where the doctors diagnosed him with Motor Neuron Disease (Spinal Muscular Atrophy). The condition was explained to the parents, and medications were prescribed. However, the parents refused to accept the diagnosis and discontinued the medications. As a result, the child's weakness progressed, leading to muscle wasting in most of the upper and lower limbs, as well as wasting of respiratory muscles, resulting in apectusexcavatum deformity. Investigations such as Nerve conduction studies shows absent compound muscle action potential with normal sensory nerve action potential and absent F wave in all tested nerves. According to the **Spinal muscular atrophy classification scale it is diagnosed as** SPINAL muscular atrophy – Type III(Kugelberg –Welander-disease).Following the suggestion of relatives, the child was taken to SV Ayurvedic Hospital in January 2025.



Picture 3: PectusExcavatum Deformity

Clinical findings:

	Case-1	**Case-2**	**Case-3**
Age	75 years	62 years	13 years
Gender	Male	Male	Male
Built	Ectomorphic	Mesomorphic	Ectomorphic
Facial asymmetry	Bilateral weakness (right>left)	Left side weakness	Not present
Skin changes	Generalized muscle wasting involving bilateral thenar and hypothenar eminences, interossei, biceps, triceps, forearm, quadriceps, hamstrings, and calf muscles	Generalized muscle wasting more pronounced on the left side (left > right), involving the bilateral thenar, hypothenar, interossei, biceps, triceps, forearm, quadriceps, hamstrings, and calf muscles	Generalized muscle wasting involving both upper and lower limbs and the trunk
Fasciculations	Tongue and upper limb muscles.	Whole body and tongue.	Whole body
Speech	Mild dysarthria	Mild dysarthria	Nasal speech
Cranial nerve examination	VII, IX, X and XII cranial nerves affected	VII, X and XII cranial nerves affected	X and XII cranial nerves affected
Sensory examination	Normal sensations	Normal sensations	Normal sensations
Deformities	Ulnar claw hand deformity of right hand	Total claw hand deformity of left hand	Pectus excavatum deformity
Pathological reflexes	Babinski sign: +Hoffmann sign: +	Babinski sign: +	Babinski sign: +

Table 1: Summary of Clinical Findings in the Three Cases

Physiotherapy interventions

Physiotherapy interventions for MNDs target respiratory difficulties, muscle weakness, and mobility impairments. Therapists concentrate on creating customised exercise regimens according to the problems list. In respiratory care, physiotherapists implement techniques to enhance lung function and guide respiratory exercises. To enhance general well-being and day-to-day functioning, the holistic approach consists of lifestyle modifications, patient education on adaptive techniques, and emotional support.

Component affected	Goal	Physiotherapy treatment
Speech and swallowing	To improve Speech and swallowing	Speech therapy, swallow exercises, adaptive strategies.
Respiratory muscles	To improve respiratory function.	Breathing exercises, respiratory muscle training, assisted cough techniques.
Facial muscles	Maintain or improve facial muscle strength	Facial exercises like cheek puffing, smile and frown exercise
Motor impairement	To improve or maintain voluntary muscle strength and function.	Range of motion exercises for both UL's and LL's, strengthening exercises and Gripping exercises
Posture and mobility	Improve or maintain mobility and posture	Passive stretching, active stretching, and gait training
Balance and coordination	Improve or maintain Balance and coordination.	Balance training,functional strength training, isometric exercises and coordination exercises.
ADLs	To improve ADLs	ADL's training
Emotionalwell-being	Support emotional health and coping strategies	Counselling, support groups, mindfulness techniques, Jacobson relaxation exercises

Follow-up and outcome measures:

Motor examination before and after treatment shows

	Case 1		**Case 2**		**Case 3**	
	Pre treatment	**Post treatment**	**Pre treatment**	**Post treatment**	**Pre treatment**	**Post treatment**
MMT						
Upper limb	2/5	4/5	3/5	4/5	3/5	3/5
Lower limb	3/5	4/5	2/5	4/5	2/5	3/5
Reflexes						
Biceps jerk	3+	2+	3+	2+	0	1+
Triceps jerk	2+	2+	2+	2+	0	1+
Knee jerk	3+	3+	3+	2+	0	2+
Ankle jerk	0	1+	1+	2+	0	1+
Plantar response	Extensor	Extensor	Flexor	Flexor	Extensor	Extensor
Berg balance scale	20/56	35/56	25/56	40/56	10/56	30/56
Fatigue severity scale	54/63	45/63	30/63	20/63	57/63	40/63
Northwick	24/34	14/34	17/34	10/34	27/34	20/34

park ADL scale						

Table 3:Motor Examination Outcome Measures Including Manual Muscle Testing (MMT), Reflexes, Balance, Fatigue, and Activities of Daily Living (ADLs) Evaluated Pre- and Post-Treatment

Muscle girth examination before and after treatment shows:

	Case 1		**Case 2**		**Case 3**	
	Pre treatment	**Post treatment**	**Pre treatment**	**Pre treatment**	**Post treatment**	**Pre treatment**
Mid arm circumference	23cm	25.5cm	25cm	27cm	16.5 cm	17 cm
Mid forearm circumference	18.5cm	20cm	20cm	22cm	13.5 cm	14 cm
Mid thigh circumference	36cm	36cm	36cm	36cm	29 cm	30 cm
Mid calf circumference	26cm	26cm	27cm	27cm	15 cm	15.5 cm

Table 4:Muscle girth examination before and after treatment

Results

A thorough neurological examination was conducted to identify the affected components in all three cases. Based on these findings, physiotherapy interventions were individually selected. The primary impairments were assessed both before and after treatment using standardized outcome measures: Manual Muscle Testing (MMT) for muscle strength, the Wexler Scale for reflex responses, the Berg Balance Scale (BBS) for balance, the Fatigue Severity Scale (FSS) for fatigue, and the Northwick Park ADL Scale for activities of daily living (ADLs).

Post-treatment results demonstrated significant improvements when compared to pre-treatment assessments:

- Muscle Strength**:** Improved by 30% in Cases 1 and 2, and by 20% in Case 3.
- Reflex Responses (Wexler Scale): Normalization improved by 25% in Cases 1 and 2, and by 31.25% in Case 3.
- Balance (BBS): Improvement of 26.79% in Cases 1 and 2, and 35.71% in Case 3.
- Fatigue (FSS): Reduction in fatigue severity by 14.29% in Case 1, 15.87% in Case 2, and 26.98% in Case 3.
- Activities of Daily Living (Northwick Park ADL Scale): Improvement of 29.41% in Case 1, and 20.59% in both Cases 2 and 3.
- Muscle Girth: An increase in muscle bulk was observed, with gains of25% in Case 1, 12.5% in Case 2, and 15.5% in Case 3.

These improvements are attributed to the application of targeted physiotherapy interventions that addressed specific neuromuscular deficits in each case, particularly through the use of selective strengthening exercises and functional retraining.

Discussion:

In bulbar variant Motor Neuron Disease (MND), rehabilitation focused on maintaining function and preventing complications like joint stiffness and muscle wasting. As the disease was progressive, a personalized, goal-based physiotherapy program was essential. Treatment was tailored to each patient's specific issues, such as muscle weakness, swallowing problems, breathing problems, poor balance, or fatigue.

Respiratory physiotherapy is especially important, as breathing muscles weaken over time (5). Techniques like deep breathing, inspiratory muscle training, and assisted coughing help improve lung function and clear secretions, reducing the risk of infection (6).

Although progressive muscle weakness without pain or sensory disturbance is a hallmark symptom of Amyotrophic Lateral Sclerosis (ALS), patients often develop secondary musculoskeletal pain and discomfort over time. This typically results from reduced mobility, joint stiffness, difficulty maintaining proper positioning in bed or a wheelchair, and the development of contractures. To address these issues, physiotherapy interventions were carefully tailored to each patient's functional level and included targeted strengthening exercises to preserve muscle function, balance training to reduce fall risk, and mobility exercises to maintain joint range and improve overall movement efficiency(7).

A recent study examined symptoms reported by people with ALS (PALS) and how these symptoms are currently managed. The most common symptoms reported were fatigue (90%), muscle stiffness (84%), muscle cramps (74%), shortness of breath (66%), difficulty sleeping (60%), and pain (59%). Despite fatigue being the most common symptom, it was also the least treated. Other symptoms with low treatment rates included shortness of breath, pain, muscle stiffness, excess saliva, pseudobulbar affect, and weight loss. Many of these symptoms can be effectively managed with physical therapy, such as energy conservation for fatigue, stretching for muscle stiffness and cramps, and tailored interventions for pain depending on its cause (8).

This targeted approach led to measurable improvements in muscle strength, balance, fatigue, respiratory function, and daily activities. It highlights the value of customized physiotherapy in improving quality of life and slowing disability, even in a progressive disease like MND(9).

Conclusion:

This case series concludes that a customized physiotherapy program played a crucial role in achieving optimal functional outcomes and significantly enhancing the patient's quality of life. Tailored interventions addressed individual needs, helping to maintain independence and manage the progressive nature of the disease more effectively. This collaborative, patient-centered model highlights the importance of a multidisciplinary healthcare team in managing the complex nature of this progressive neurodegenerative condition.

Reference:

1. The Scottish Motor Neuron Disease Register: a prospective study of adult onset motor neuron disease in Scotland. Methodology, demography and clinical features of incident cases in 1989.Arora

R, Khan Y. J NeurolNeurosurg Psychiatry. 1992;55:536–541
2. Nilay M, Moirangthem A, Saxena D, et al. Carrier frequency of SMN1- related spinal muscular atrophy in north Indian population: The need for population based screening program. Am J Med Genet A. 2020. October 14.
3. Traynor BJ, Codd MB, Corr B, Forde C, Frost E, Hardiman OM. Clinical features of amyotrophic lateral sclerosis according to the El Escorial and Airlie House diagnostic criteria: A population-based study. Arch Neurol. 2000 Aug;57(8):1171-6.
4. Ingre C, Roos PM, Piehl F, Kamel F, Fang F. Risk factors for amyotrophic lateral sclerosis. ClinEpidemiol. 2015;7:181-93.
5. Desnuelle, C., Bruno, M., Soriani, M. H., & Perrin, C. (2006). Quellessont les modalités de thérapie physique symptomatiqueincluant les techniques de désencombrementbronchique? [What physical therapy techniques can be used to improve airway freedom in amyotrophic lateral sclerosis?]. *Revue neurologique*, *162 Spec No 2*, 4S244–4S252.
6. Lacombe M, Del AmoCastrillo L, Boré A, Chapeau D, Horvat E, Vaugier I, Lejaille M, Orlikowski D, Prigent H, Lofaso F. Comparison of three cough-augmentation techniques in neuromuscular patients: mechanical insufflation combined with manually assisted cough, insufflation-exsufflation alone and insufflation-exsufflation combined with manually assisted cough. Respiration. 2014;88(3):215-22.
7. Ashworth, N. L., Satkunam, L. E., &Deforge, D. (2004). Treatment for spasticity in amyotrophic lateral sclerosis/motor neuron disease. *The Cochrane database of systematic reviews*, (1), CD004156.
8. Nicholson K, Murphy A, Mcdonnell E, et al. Improving symptom management for people with amyotrophic lateral sclerosis. Muscle Nerve. 2017;57(1):20–24.
9. 9.Bello-Haas VD. Physical therapy for individuals with amyotrophic lateral sclerosis: current insights. DegenerNeurolNeuromuscul Dis. 2018;8:45-54.