Case Report on the role of physiotherapy in improving the quality of life of a patient with Amyotrophic lateral sclerosis

Tejal Babar, Ragini Dadgal, Rakesh Kovela, Moh’d Irshad Qureshi

ABSTRACT

The commonest type of motor neuron disease is ALS. Amyotrophic lateral sclerosis (ALS), which is also known as Lou Gehrig’s disease, is a degenerative disease that affects the neurons and includes features of gradually increasing motor neurons degeneration that controls voluntary muscles functions. Early signs of ALS include gradual onset of stiffness of muscles, muscular spasms, progressive weakness, and muscle wasting. C9orf72 (chromosome 9 open reading frame 72) is the commonest gene that undergoes mutation in ALS, promoting the loss of motor neurons in multiple ways. This case is of a 47 years old male patient who presented with complaints of asthenia in the lower limb for 2 years, asthenia in the upper limbs for 1½ years, dyspnea for the last 5 months, and dysphasia, dysphagia, sialorrhea for the last 3 months. He underwent various interventions like MRI, CT scans, EMG (Electromyography), and NCV (Nerve Conduction Velocity). He was then diagnosed with Motor neuron disease after ruling out another probable diagnosis like upper and lower motor neuron disease, Syringomyelia. The disease-specific outcome measures for ALS are ALSFRS and ALSAQ-5. Physiotherapy treatment that is modulated according to the patient’s need plays a vital role in improving the quality of life and helps in delaying the worsening of symptoms henceforth helping in increasing the life span of the patients diagnosed with Amyotrophic Lateral Sclerosis.

Keywords: Amyotrophic lateral sclerosis, Neurodegeneration, Sporadic, Muscle Twitches, Gene Mutation.

1. INTRODUCTION

Amyotrophic lateral sclerosis (ALS) is an unusual neurological illness that is deadly. The illness is distinguished by the motor neurons degeneration in the cortical regions along with the brain stem and spinal cord, resulting in motor deficits and paralysis of the muscles in a progressive manner regulating the movement of limbs, swallowing, and breathing (Meyer et al., 2018). Upper and lower motor neurons degeneration in the motor cortical region, nuclei of the brain stem, and the anterior horn cells of the spinal cord led to gradual
weakness and wasting of muscles. ALS frequently begins locally, but then affects multiple bodily parts, with loss of function of the muscles responsible for respiratory, generally limiting the patient’s life span to 2-5 years following illness onset (Marsro and Van Damme, 2020). Because of the intricacy of the central and peripheral neurological systems, ALS symptoms include muscular atrophy, weakness, stiffness, exhaustion, trouble speaking, difficulty swallowing, and shortness of breath. The exorbitant sign of ALS is considered to be Asthenia (Park et al., 2020).

The relationship between the features and the underlying pathology was originally reported in 1869 by a neurologist named Jean-Martin Charcot, a French Physician who gave this name amyotrophic lateral sclerosis during the 1870s (Rowland, 2001). It came into the limelight in the US when it was diagnosed by baseball star Lou Gehrig in 1939, and then internationally as cosmologist Stephen Hawking was identified with this disease in 1963. People with ALS having an early-onset are usually male, having bulbar onset and a slow progression of the disease. The Sensory nerves along with the autonomic nervous system stay largely untouched. Asthenia is the first indication of ALS. Other features include dysphagia, cramps or stiffness of involved muscles, asthenia in arms or legs, and slurred speech. Early ALS symptoms impact different regions of the body that solely is based on specific motor neurons that are initially injured. The initial signs of limb-onset ALS are either in the upper or lower limbs. If the limb onset is present, the patient will be suffering discomfort and stumbling while walking or jogging; which is commonly characterized by a gait with a “foot drop” dragging lightly over the floor. If limb onset is present, individuals will be having difficulty with manual dexterity activities like buttoning-unbuttoning a shirt, holding a pen for writing, twisting the keys of a car, and locking (Dharmadasa et al., 2018).

People gradually lose their ability to move, swallow (dysphagia), talk, or make words (dysarthria). Twitching, on the other hand, is not more of a diagnostic sign but a side effect; occurring after or in conjunction with asthenia. Jason Becker who was a guitarist had the condition since 1989, while on the other hand Stephen Hawking a famous cosmologist survived another 55 years after being diagnosed, although both are regarded as outliers. Environmental toxins (as determined by regional deployment studies). A review in 2016 of 16 meta-analyses revealed that there was affirmation for an alliance with severe occupational lead exposure; beta-carotene intake, poor proof of intake with an omega-3 fatty acid, vulnerability to utmost low-frequency electromagnetic fields and pesticides (Belbasis et al., 2016). The physical pathology of ALS includes skeletal muscle atrophy, motor cortical atrophy, dwindling of the hypoglossal nerves (that control the tongue), and the anterior roots of the spinal cord. Aside from motor neuron death, two other features shared by most ALS variations are localized initial pathology, which means that symptoms begin in a particular spinal cord location, and developing uninterrupted expansion, which means that symptoms extend to further regions over time. Motor neurons have more chances of excitotoxicity compared to the rest of the neurons due to a reduced capacity of calcium-buffering and a glutamate receptor which has more permeability to calcium. Riluzole, a drug that has been found to delay deterioration in ALS, inhibits the release of glutamate from presynaptic neurons; although, whether the mechanism is in charge of its therapeutic effect or not is unclear (Grad et al., 2017).

2. PATIENT INFORMATION

This case is of a 47 years old male patient who was a Guard supervisor by occupation with right-handed dominance residing in Nagpur. The informant was the patient’s wife. He was presented with the complaints of asthenia in the lower limb for 2years, asthenia in the upper limbs sifor½ years, dyspnea for the last 5 months, and dysphasia, dysphagia, sialorrhea for the last 3 months. The patient was apparently alright 2½ years back when he started feeling low back pain. The pain was insidious in onset with no diurnal variation and was hampering his day-to-day activities. After about 6 months, he started experiencing weakness in bilateral lower limbs during weight-bearing which was gradually progressing over time and he had to cease his job. For these complaints, he visited a private clinic in Nagpur on 15 October 2019 and was given certain medications. Then after about 6 months of the onset of lower limb symptoms, he started experiencing upper limb weakness as he was having difficulty in performing activities like brushing his teeth, combing his hair, holding a newspaper, and turning pages. For these complaints, he visited the same clinic that he previously did and was told to undergo certain investigations. In those investigations, it was divulged that he is suffering from Amyotrophic Lateral Sclerosis. He was referred to a multi-specialty hospital in Nagpur and admitted for 20 days. He also has had complaints of occasional dry cough during supine position for the last 3 months. This symptom was usually neglected by the family and caretakers of the patient as the symptoms improved as he changed his position from supine to sitting. He visited AVBHRH (Acharya Vinobha Bhave Rural Hospital) on 25/12/2021 for his presenting complaints and was referred for physiotherapy the next day. He has a history of alcohol consumption of 24 years along with smoking for the last 17 years but has ceased since February 2020. He is a k/c/o Hyperlipidemia for the last 8months with no other co-morbidities. His socioeconomic status was revealed as Upper middle class on the Modified Kuppuswamy scale.
3. CLINICAL FINDINGS

The patient was assessed in a supine lying position on the couch with a pillow under the cervical spine for support. The vital signs like heart rate were increased to 109 beats/min; respiratory rate was increased to 29 breaths/min. On observation, the patient had an ectomorphic build. The attitude of limbs for the upper extremity of the right side was horizontal adduction of the shoulder with elbow and wrist in flexion towards his chest while on the left side; his shoulder was abducted, internally rotated with elbows and wrist in extension by the side. In bilateral lower limbs, his hips were extended, adducted, and externally rotated with the knee in extension in slight Plantarflexion. There was the use of accessory muscles of respiration with the thoracoabdominal type of breathing due to weakness of the diaphragm and muscles of respiration. There was no evidence of edema or pressure sores. On Palpation, there were no noteworthy findings in the context of warmth, tenderness, and swelling.

On examination, the higher mental functions were normal i.e. the cognition of the patient was not impaired as the score was 28/30 on the Mini-mental scale examination (MMSE). His short- and long-term memory was intact. In special senses, V (Motor part) (VII (Motor part), IX (motor part), X, XI & XII (motor part) cranial nerves were impaired. In the sensory examination, the superficial, deep, and cortical sensations were intact bilaterally over the upper and lower limbs along with the trunk. In the motor examination, his muscle atrophy was evident over superficial flexor compartment muscles of the forearm- Flexor carpi radialis, Flexor carpi ulnaris & Pronator teres, Tibialis anterior, Gastrocnemius, and Soleus as the muscles were flat and undernourished in the appearance of bilateral lower limbs. On voluntary control grading, his upper limbs had a grade of 4 and his lower limbs had a grade of 1. Muscle tone according to the Modified Ashworth Scale of upper limb was 1+ in the bilateral shoulder, elbow, and wrist joints. His lower limbs were flaccid. His deep tendon reflexes grading is given in Table 1.

Table 1 Grading of Deep Tendon Reflexes.

<table>
<thead>
<tr>
<th>DEEP TENDON REFLEX</th>
<th>GRADEING</th>
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<tr>
<td>Jaw jerk</td>
<td>3+ (Brisker or more reflexive than normal)</td>
</tr>
<tr>
<td>Biceps jerk</td>
<td>3+</td>
</tr>
<tr>
<td>Supinater jerk</td>
<td>2+ (Normal)</td>
</tr>
<tr>
<td>Triceps jerk</td>
<td>3+</td>
</tr>
<tr>
<td>Knee jerk</td>
<td>1+ (Low normal, diminished)</td>
</tr>
<tr>
<td>Ankle jerk</td>
<td>0 (Absent, no response)</td>
</tr>
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In the integumentary system, skin status was dry and flaky. On inspection, in respiratory assessment, the anteroposterior diameter was less than the transverse diameter. Flattening of the chest was observed. His shoulders were depressed. The respiratory rate was 29 breaths/min. The utilization of accessory muscles of respiration like sternocleidomastoid & scalene during breathing was seen, the character was Abdominothoracic due to weakness of the diaphragm and other respiratory muscles.

Timeline

October 2019: Lumbar spine MRI was done. 3rd April 2020: CBC (Complete Blood Count), ESR (Erythrocyte Sedimentation Rate), Urine Analysis, Lipid profile test, Kidney function test, Kidney basic screening, Random glucose, Immunology, Serology - (HbsAg, Rapid plasma reagin), LFT (Liver Function Test), Thyroid profile test, Vitamin B12, Calcium. 16th April 2020: MRI of the whole spine. 17th April 2020: Diagnosed with Motor Neuron Disease- Amyotrophic Lateral Sclerosis. 20th April 2020: Cervical spine MRI. 21st April 2020: EMG, NCV that concluded anterior horn cell lesion. 26th April 2020: MRI (Magnetic Resonance Imaging) of the brain & screening of the whole spine.

Diagnostic Assessment

Diagnostic methods

The patient underwent an investigation of a lumbar spine MRI in October 2019 which revealed small size posteroventral disc protrusions at L2-3, L4-5 levels causing an indentation in thecal sac. He then underwent further investigations like CBC that revealed increased RBC count-5.45 mil/mL, ESR, Urine Analysis, Lipid profile test, Kidney function test, Kidney basic screening that revealed an elevated level of uric acid-6.0 mg/dL, Random glucose, Immunology, Serology for Hbs Ag, HIV, Rapid plasma reagin, LFT (Liver Function Test) that revealed an elevated level of triglycerides-176mg/dL and Alanine Transaminase (ALT) – 35 U/L, Thyroid profile test, Vitamin B12, Calcium on 03/04/2021. On 16/04/2020, he underwent an MRI of the whole spine which divulged a posterior disc bulge at the cervical and lumbar levels. Then he was diagnosed with Motor neuron Disease. On 20/04/2021, he
underwent a cervical spine MRI. On 21/04/2020, NCV and EMG were carried out: Motor nerve conduction showed slightly reduced Compound Motor action potential (CMAP) amplitude from the right ulnar nerve and right peroneal nerve.

NEE (Needle examination) showed increased insertion activity with widespread fibrillation potentials, and positive waves from distal and proximal muscles of all four limbs, cervical, thoracic, and lumbosacral paraspinal mentalis. Long duration, high amplitude Motor unit action potentials (MUAPs) was seen with reduced recruitment pattern from proximal and distal muscles of upper and lower limbs bilaterally. The conclusion was widespread AHC lesion with active chronic denervation in muscles involving lumbosacral, thoracic, and cervical segments and so he was also diagnosed with an anterior horn cell lesion. On 26/12/2021, he underwent a few more investigations like a Brain MRI that had the impression of mild cerebral and cerebellar atrophy and screening of the whole spine that divulged findings of early degenerative spondylotic changes in the spine with marginal endplate osteophytes and desiccated intervertebral discs that displayed hypointense signal on T2 weighted images at all cervical and 2-3 lumbar level. Posterior disc bulge was noted at C3-4 to C5-6 levels indenting the thecal sac without overt compression of nerve root without notice of any compressive or non-compressive myelopathy. The posterior disc bulge was divulged at the L2-3 level without nerve roots compression (Figure 1).

![MRI of the brain.](image)

**Figure 1** MRI of the brain.

**Diagnostic Challenges**
Disease heterogeneity and rarity, fast progression, lack of etiology understanding, and lack of biomarkers are the few diagnostic challenges.

**Diagnosis**
As this is a complex illness intertwining along with a variety of different disorders, diagnosis at the initial phase is challenging, he was diagnosed with Amyotrophic lateral sclerosis by ruling out all the other possible diagnoses.
Therapeutic Interventions
Owing to the individual variability of the disease onset, disease course, and disease progression, patients with ALS will present with unique and different sets of problems thus, the interventions will vary. Goal framing in ALS has been such that the physiotherapist along with the patient knows the things to anticipate from treatment. The goals focus to maintain the function makes the patients independent. We'll give a directed strategy that is subject to adjustment as the condition advances by identifying targets.

The goals mainly focus: To preserve and improve mobility and function; to address any signs of respiratory impairment; in improving the patient’s quality of life; to avoid impairment in strength or mobility due to the condition’s progressive nature; to prevent subsequent musculoskeletal injuries by providing hoists or manual handling belts early on; Treatment proceeded with the patient and relative education about his condition and the importance of physiotherapy for his prognosis (Figure 2).

Figure 2 patient treatments in Physiotherapy OPD

Phase I (Day 1 to 4 weeks)
Stretching of Gastrocnemius-Soleus, Hamstrings, Adductors, Flexor carpi ulnaris and radialis, Biceps Brachii, Trapezius, Scalene muscles with 3 repetitions with 30 seconds hold in 1 set. Passive movements to Toes, Ankle, knee, and hip joints with 20 repetitions in 1 set; Active Assisted Range of movements to fingers, wrist, elbow, and shoulder joints with 10 repetitions in 1 set.

Rood’s Approach
Facilitatory techniques for lower limbs like fast brushing, quick icing on muscle belly, tapping, and heavy joint compression to treat flaccidity. Inhibitory techniques for upper limb like slow stroking and rolling, prolonged stretch, pressure on insertion of the muscle, light joint compression, resisted contractions to treat spasticity. Breathing Exercises like pursed-lip breathing and glossopharyngeal breathing in intervals between each set of exercises and advised practicing as many times possible throughout the day. Bed Mobility exercises were taught to the patient for supine to side-lying like log-rolling and segmental rolling with assistance. Advise on the avoidance of citrus fruits and alcohol.

Oral facial facilitation to aid motor control, sensory awareness, and swallowing frequency control; Icing over platysma and facial muscles for 5-20 minutes to improve tone and swallow reflex. Brushing for 15-20 minutes is undertaken before meals. Vibration to improve tone; Manipulation techniques such as tapping, stroking, patting, and applying strong pressure directly to muscles with the fingertips can help enhance oral awareness. Oral motor sensory exercises like lip closure, tongue movement, and deglutition to improve chewing and slurring skills; Skin hygiene like ensuring sponge bath of patient twice daily followed by proper wiping with a clean towel or cloth; Application of humectant-based moisturizer or lotion to avoid dryness of the skin.

Phase II (4 to 8 Weeks)
Stretching of Gastrocnemius-Soleus, Hamstrings, Adductors, Flexor carpi ulnaris and radialis, Biceps Brachii, Trapezius, Scalene muscles with 3 repetitions with 30 seconds hold in 1 set. Passive movements to Toes, Ankle, knee, and hip joints with 20 repetitions.
in 1 set twice a day. Active Assisted Range of movements to fingers, wrist, elbow, and shoulder joints with 20 repetitions in 1 set twice a day.

**Roods Approach**
Facilitatory techniques for lower limbs like fast brushing, quick icing on muscle belly, tapping, and heavy joint compression to treat flaccidity. Inhibitory techniques for upper limb like slow stroking and rolling, prolonged stretch, pressure on insertion of the muscle, light joint compression, unresisted contractions to treat spasticity. Breathing Exercises like pursed-lip breathing and glossopharyngeal breathing in intervals between each set of exercises and advised practicing as many times possible throughout the day. Bed Mobility exercises were taught to the patient for supine to side-lying with segmental rolling. Oral facial facilitation to aid motor control, sensory awareness, and swallowing frequency control; Icing over platysma and facial muscles for 15-25 minutes to improve tone and swallow reflex. Brushing for 10 minutes is undertaken before meals. Vibration to improve tone; Manipulation like tapping, stroking, patting, and firm pressure directly over muscles using fingertips to improve oral awareness. Oral motor sensory exercises like lip closure, tongue movement, and deglutition to improve chewing and slurring skills; Application of humectant-based moisturizer or lotion to avoid dryness of the skin.

**Outcomes of interventions**
The ALS Functional Rating Scale (revised) - ALSFRS-R examines the functional status of patients. This scale is arranged in ascending manner i.e it scores from 4 (normal function) to 0 (unable to attempt function) (Figure 3).

![ALSFRS-R](image)

**Figure 3** ALSFRS-R examines the functional status of patients.

The ALS Assessment Questionnaire (ALSAQ-5) is a disease-specific quality of life measure for ALS that contains 5 items that measure health status in the last 1 week in each domain using a summary score from 0- never, 1- rarely, 2- sometimes, 3- often, 4-always. The lowest score depicts a good prognosis while the higher score depicts the severity of the disease (Figure 4).
4. DISCUSSION

Booth et al., (2012) stated the benefits of exercise in healthy adults are well known and well-proven. Similarly, physical inactivity is related to a greater threat related to occurrence in a wide range of uncertain health-related issues. Indeed, at the single motor unit level, age along with co-morbidities has a proportional linkage without muscle atrophy. Moreover, current research indicates that exercise improves muscle fiber reinnervation in the elderly. Along with advantages, they discovered comprehensive refinement in the scoring of the functional independence scale, regardless of the category of physical activity performed. Furthermore, gains occurring in muscular power, oxygen consumption, and tiredness had been notably found in the group of EP (exercise program), all of which are characteristics of the results of physical activity for the particular exercise regime (Booth et al., 2012).

Pinto et al., (1999) studied the aspect of slow motor degeneration in ALS patients after prescribing exercise to them to the level of anaerobic threshold ensuring respiratory insufficiency by using the Bipap STD®, a non-invasive ventilator. When compared to the non-exercise control group, ALS patients who followed an exercise routine on the treadmill for 10-11 months had a lower incidence of reduction of respiratory capacities and a greater scoring of functional independent mobility (Pinto et al., 1999). Accordingly, Sanjak et al., (2010) showed 30 minutes of treadmill thrice a week for 2 months profoundly led to improvement in the scoring of the ALS Functional Rating Scale (ALSFRS) along with tolerability, gait speed, distance, and stride length during 6-minute walk tests for the same population (Sanjak et al., 2010).

Cheah et al., (2009) told that the ability to venture endurance training at a moderate pace with non-invasive ventilation or a bodyweight supporting device in ALS patients was lately manifested, with advancing functions and cardiorespiratory capacities, suggesting that endurance training done at a home setup can protect ALS patients from the repercussion that occur after progressive degenerative changes. Lately revealed 24-week endurance training with a mini-cycle as non-tolerable compared to resistance training or stretching exercises for the upper and lower body. Considering it not being in the focus, the potential of respiratory training is worth noticing for its role in enhancing its function, considering it as the primary aspect of aerobic or endurance training, in patients with ALS. There is also the evidence of Scientific research suggesting a 12-week inspiratory muscle training program consisting of breathing and exhaling using a particular device (Respironics®) leads to a pause or slowing down in the pace of deterioration in respiratory function in ALS patients by strengthening their muscles of inspiration (Cheah et al., 2009).

5. CONCLUSION

Although the prognosis of ALS is poor and the quality of life of the patient is drastically hampered, physiotherapy treatment that is individualized to the patient’s requirements and objectives and which focuses on managing symptoms, enhancing function and involvement helps persons with ALS to live their lives with quality.
Acknowledgment
We hereby thank all the authors and guides for their contribution in the form of immense support, tutoring, and briefing for the preparedness of this case report.

Author’s Contributions
TB and RD evaluated the case report, and RD & RK helped in the final drafting of the case report. TB, RD, RK, and IQ assisted in the revision of the manuscript. All authors’ approved the final version of the manuscript.

Patient’s Perspective
After 4 weeks of my physiotherapy treatment, I’ve started feeling much better, reduce my dyspnea significantly, and it also indulged a sense of motivation in me to be active as much as possible and taught me to always be willing for exercise sessions, which I was unable to do before the physiotherapy treatment.

Authors’ contributions
The design of this report was suggested by TKB and RD. The study was made and implemented with the help of TKB and RD. TKB wrote this report. The final report was analyzed and approved for publishing by RD.

Informed Consent
The patient was first informed about the study followed by obtaining oral informed consent.

Funding
This study has not received any external funding.

Conflicts of interest
The authors declare that there are no conflicts of interests.

Data and materials availability
All data associated with this study are present in the paper.

REFERENCES AND NOTES