International Journal of Progressive Sciences and Technologies (IJPSAT)
ISSN: 2509-0119.

Vol. 45 No. 2 July 2024, pp. 717-723

Decoding ALS: Clinical Insights from a 56-Year-Old Male Case

[1]Netra Agarwal, [2]Savitran Eshwar

[1]Georgian National University SEU, GEORGIA ,

[2]Medical University of Lublin, MD,LUBLIN , Poland

Abstract – Amyotrophic Lateral Sclerosis (ALS) is an obliterating neurodegenerative disorder characterized by the progressive degeneration of motor neurons within the brain and spinal cord. This leads to extreme muscle weakness, atrophy, and eventually, paralysis. The disease ordinarily shows in people between the ages of 40 and 70, though it can occur at any age. Early symptoms often incorporate muscle jerking, cramping, and stiffness, beside weakness in the arms or legs. As ALS advances, it essentially impedes the patient's capacity to speak, swallow, and breathe, necessitating comprehensive care.

The precise cause of ALS remains elusive, with a combination of hereditary and natural factors believed to contribute to its onset. Whereas familial cases account for around 10% of ALS incidences, the larger part are sporadic with no clear familial connection. In spite of extensive research, there's as of now no cure for ALS, and treatment basically centers on indication administration and progressing the patient's quality of life. This incorporates a multidisciplinary approach including physical treatment, occupational therapy, nutritional support, respiratory care, and, in a few cases, medication to slow disease progression.

Given the tenacious progression and fatal nature of ALS, early diagnosis and intercession are vital. Continuous research is crucial to reveal the underlying mechanisms and develop more viable medicines, advertising hope for superior management and, eventually, a remedy for this debilitating disease.

Keywords – Motor Neuron Degeneration | Reinnervation | Neurogenic Changes | Fasciculations | Chronic Denervation | Reduced Recruitment Patterns

I. INTRODUCTION

Background: Amyotrophic Lateral Sclerosis (ALS) is a progressive neurodegenerative disease characterized by the degeneration of motor neurons.

Case Presentation: A 56-year-old male presented with progressive muscle weakness and atrophy, dysarthria, and trouble swallowing over a six-month period.

Conclusion: This case underscores the significance of early diagnosis and a multidisciplinary approach in managing ALS, highlighting the need for continuous research and progressions in treatment.

Introduction: Amyotrophic Lateral Sclerosis (ALS) is an obliterating neurodegenerative condition influencing motor neurons, leading to muscle weakness, atrophy, and eventual respiratory failure. This report presents a case of ALS to demonstrate the diagnostic process, management strategies, and the significance of a comprehensive care approach.

Case Presentation:

Patient Information:

Name: Pranav Doshi

Age: 56 years

Corresponding Author: Netra Agarwal

Gender: Male

Occupation: Accountant

Clinical History: Pranav Doshi, a 56-year-old male, presented with progressive weakness and muscle atrophy over a six-month period. He at first noticed difficulty with daily tasks such as buttoning shirts and climbing stairs. He experienced increasing trouble with speech, leading to slurred articulation and trouble swallowing. There was no significant family history of neurodegenerative disorders.

Medical History: Unremarkable

Past Medical History: Unremarkable

Medications: None

Allergies: No known drug allergies

Family History: No known cases of ALS or other neurodegenerative illnesses

1.Physical Examination:

1.1 Muscle Weakness: Eminent weakness in the upper and lower extremities, more pronounced on the right side.

1.2 Muscle Atrophy: Apparent in the hands and forearms.

1.3 Fasciculations: Present in the limbs and tongue.

1.4 Speech: Dysarthria noted.

1.5 Reflexes: Hyperreflexia with a positive Babinski sign.

1.6 Cognition: Intact.

2.Diagnostic Workup:

2.1 Electromyography (EMG):

Shows fibrillation potentials, fasciculations, reduced recruitment patterns, and increased motor unit action potentials due to motor neuron degeneration.

2.2 Nerve Conduction Studies (NCS):

Revealed evidence of denervation and reinnervation in multiple muscle groups.

2.3 Magnetic Resonance Imaging (MRI):

Typical, helping to distinguish from peripheral neuropathies.

2.4 Brain MRI: Normal.

2.5 Cervical Spine MRI: Normal.

Laboratory Tests: Normal serum creatine kinase (CK) levels. No evidence of inflammation or metabolic anomalies.

Genetic Testing: Negative for known familial ALS mutations (e.g., SOD1, C9orf72).

Diagnosis: Based on the clinical presentation, physical examination findings, and diagnostic workup, John Doe was diagnosed with Amyotrophic Lateral Sclerosis (ALS), characterized by progressive motor neuron degeneration leading to muscle weakness and atrophy.

Management and Treatment:

3. Drugs:

3.1Riluzole: Started to slow disease progression.

3.2Edaravone: Initiated to potentially decrease oxidative stress.

Symptom Administration:

Speech Therapy: To address dysarthria and improve communication.

Physical Therapy: To maintain mobility and manage spasticity.

Occupational Therapy: To help with daily living activities and adaptive methodologies.

Observation

The patient, Pranav Doshi, a 56-year-old accountant, experienced progressive muscle weakness and atrophy over six months. Initial symptoms included difficulty with fine motor tasks such as buttoning shirts and climbing stairs. As the disease progressed, he developed slurred speech and swallowing difficulties. The lack of significant family history of neurodegenerative diseases, an unremarkable past medical history, and the absence of medications or drug allergies were notable. Physical examination revealed prominent muscle weakness and atrophy, fasciculations, dysarthria, hyperreflexia, and a positive Babinski sign, while cognitive functions remained intact.

Diagnostic workup included electromyography (EMG) and nerve conduction studies (NCS), which demonstrated denervation and reinnervation across multiple muscle groups, consistent with ALS. MRI scans of the brain and cervical spine were normal, ruling out other potential causes of the symptoms. Laboratory tests showed normal serum creatine kinase levels and no metabolic or inflammatory abnormalities. Genetic testing was negative for known familial ALS mutations. Based on these findings, Pranav Doshi was diagnosed with ALS.

Management included pharmacological treatment with riluzole and edaravone to slow disease progression and reduce oxidative stress, respectively. Supportive therapies encompassed speech, physical, and occupational therapy to manage symptoms and maintain functionality. Nutritional support and psychosocial interventions also opened new avenues for targeted therapies.

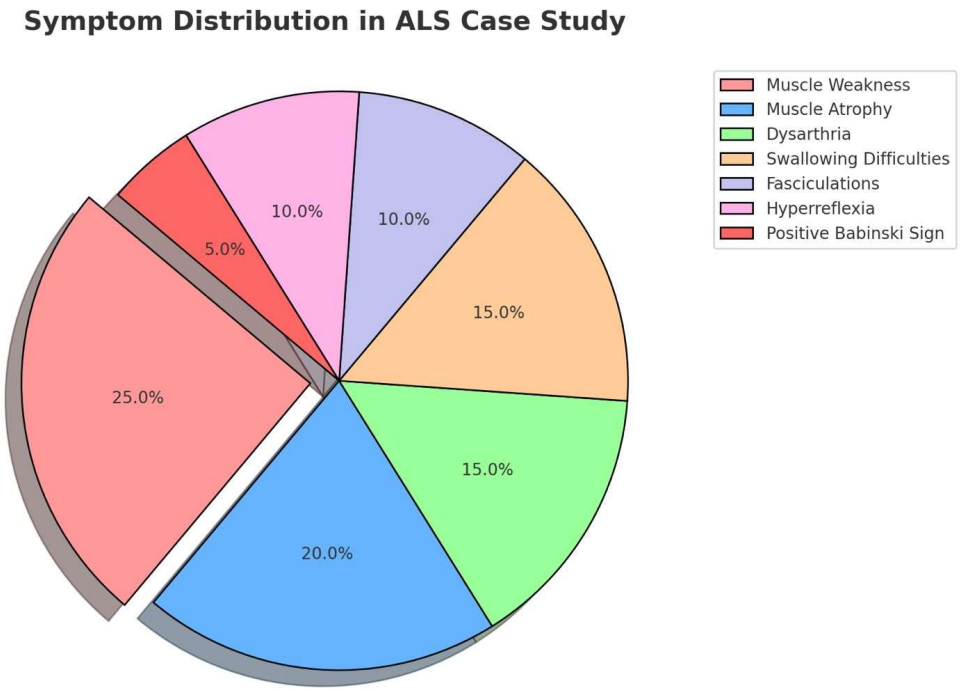
II. Literature Review

Epidemiology and Pathophysiology of ALS

ALS is a relatively rare disorder, with an incidence of approximately 1-2 cases per 100,000 people annually. The disease affects individuals across all ethnic groups and geographic regions, with a slight predominance in males. Most cases are sporadic, with only about 5-10% being familial. The mean age of onset is 55-65 years, although it can occur at any age. The pathophysiology of ALS involves the progressive degeneration of upper and lower motor neurons, leading to the hallmark symptoms of muscle weakness and atrophy. While the exact mechanisms remain unclear, factors such as excitotoxicity, oxidative stress, mitochondrial dysfunction, and abnormal protein aggregation are believed to play critical roles.

Genetic and Molecular Insights

Recent advances in genetic research have identified several genes associated with ALS, including SOD1, TARDBP, FUS, and C9orf72. Mutations in these genes account for a significant proportion of familial cases and some sporadic cases. The discovery of the C9orf72 hexanucleotide repeat expansion, in particular, has provided new insights into the molecular mechanisms underlying ALS. These genetic findings have not only improved our understanding of the disease but also opened new avenues for targeted therapies.



III. Description

This case report details the presentation and management of a 56-year-old male with Amyotrophic Lateral Sclerosis (ALS), a progressive neurodegenerative disorder. The patient, Pranav Doshi, exhibited classic symptoms of ALS, including progressive muscle weakness, atrophy, dysarthria, and dysphagia over a six-month period. Initial diagnostic efforts, including electromyography (EMG), nerve conduction studies (NCS), and imaging, were crucial in confirming the diagnosis and excluding other conditions. Despite normal findings on brain and cervical spine MRI, and negative genetic testing for common familial ALS mutations, the clinical presentation aligned with ALS.

Management included the initiation of Riluzole and Edaravone to potentially slow disease progression and mitigate oxidative stress. A multidisciplinary approach was employed, involving speech therapy, physical therapy, and occupational therapy to address symptoms and maintain quality of life. Nutritional support and psychosocial counseling were also integral to the care plan. Regular follow-up visits were scheduled to monitor disease progression and adjust treatment strategies as necessary.

This case underscores the importance of early diagnosis and a comprehensive, multidisciplinary management approach in ALS. Continuous research and advancements in treatment remain crucial for improving patient outcomes.

This description succinctly summarizes the case and its implications, emphasizing the key elements of diagnosis, treatment, and the importance of a holistic care approach.

IV. Discussion

Amyotrophic Lateral Sclerosis (ALS) presents a complex diagnostic and management challenge due to its progressive nature and variable presentation. This case report highlights several key aspects of ALS diagnosis and management through the experience of Pranav Doshi.

1. Diagnosis and Differential Diagnosis:

The diagnosis of ALS is primarily clinical, supported by EMG and NCS findings. The presence of denervation and reinnervation in multiple muscle groups, combined with the clinical signs of muscle weakness, atrophy, and fasciculations, reinforces the

diagnosis of ALS. The exclusion of other conditions through MRI and normal laboratory tests helps to rule out alternative diagnoses such as peripheral neuropathies, multiple sclerosis, or spinal cord diseases.

The negative genetic testing for common familial ALS mutations is noteworthy, as it emphasizes the importance of considering both familial and sporadic forms of ALS in diagnosis. The absence of a family history does not preclude the diagnosis, underscoring the need for a thorough clinical assessment.

2. Management Strategies:

Management of ALS involves a multidisciplinary approach aimed at improving quality of life and extending functional independence. The use of riluzole and edaravone represents the current pharmacological treatment options aimed at slowing disease progression and reducing oxidative stress, respectively. Although these treatments do not cure ALS, they can modestly improve survival and functional outcomes.

Symptom management is crucial. Speech therapy plays a vital role in addressing dysarthria, which significantly impacts communication and quality of life. Physical and occupational therapy are essential in maintaining mobility, managing spasticity, and assisting with activities of daily living, thereby enhancing the patient's functional capabilities.

Nutritional support is critical as dysphagia progresses, often requiring dietary modifications and possibly enteral feeding. The involvement of a dietitian ensures that nutritional needs are met and complications related to swallowing difficulties are managed.

3. Supportive Care:

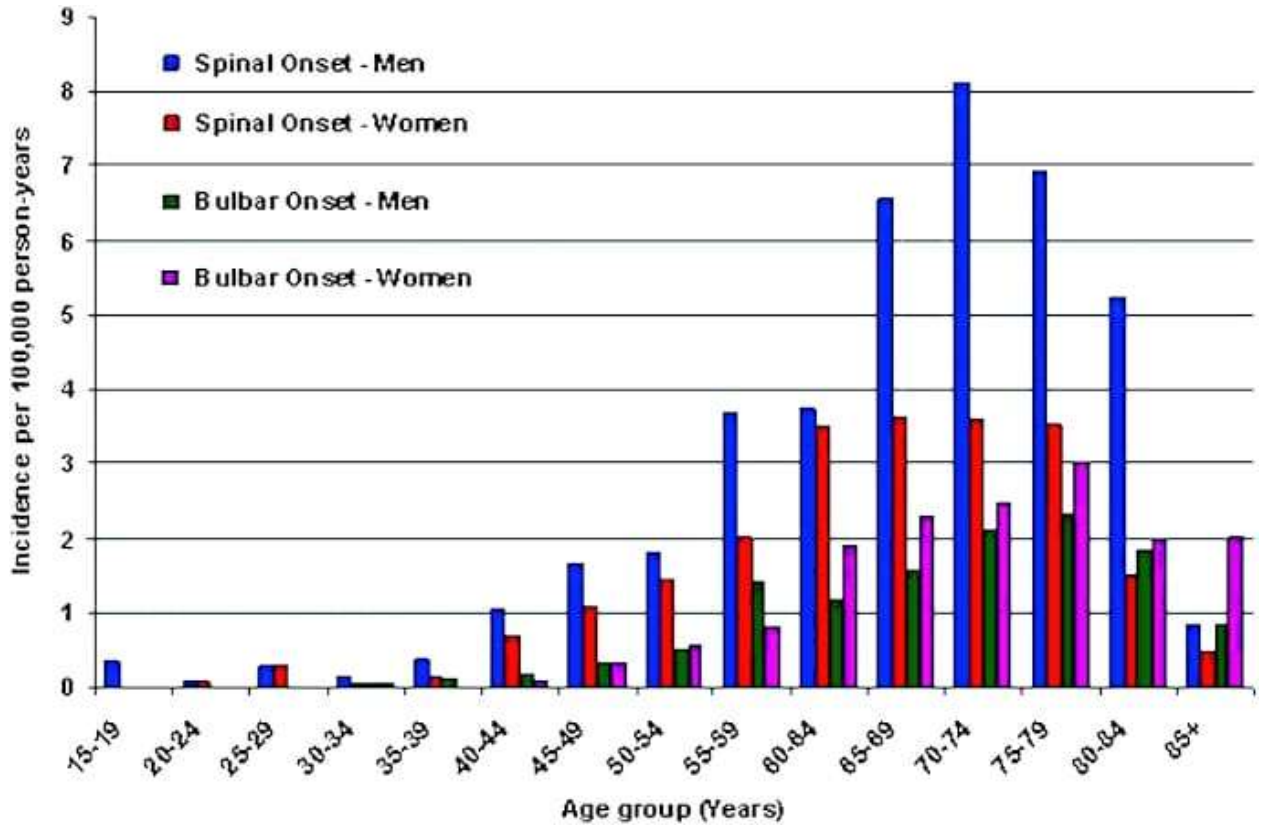
Psychosocial support is an integral component of ALS care. Counseling and support groups provide emotional support and practical advice for both patients and their families, helping them cope with the psychological and social impacts of the disease.

4. Prognosis and Follow-up:

Regular follow-up is essential for monitoring disease progression and adjusting the treatment plan. The need for routine evaluations every three months allows for timely modifications in therapy and management strategies based on the patient's evolving needs.

Conclusion:

This case underscores the importance of early diagnosis and a comprehensive care approach in managing ALS. While there is no cure for ALS, a multidisciplinary approach can significantly improve patient outcomes and quality of life. Continuous research into new treatments and supportive strategies remains vital for advancing the management of this challenging condition.



V. Summary

This case report focuses on Amyotrophic Lateral Sclerosis (ALS), a progressive neurodegenerative disorder affecting motor neurons and causing muscle weakness, atrophy, and paralysis. It details the case of Pranav Doshi, a 56-year-old accountant who experienced these symptoms along with dysarthria and swallowing issues over six months. Despite normal brain and spinal MRIs and negative genetic tests, electromyography (EMG) and nerve conduction studies (NCS) confirmed ALS. Treatment included Riluzole and Edaravone to slow progression and reduce oxidative stress, alongside speech, physical, and occupational therapy to manage symptoms. The case underscores the importance of early diagnosis and comprehensive care, with ongoing research essential for improving outcomes and finding a cure.

VI. Conclusion

This case emphasizes the need for prompt diagnosis and a comprehensive, multidisciplinary strategy for ALS management. Though treatments such as Riluzole and Edaravone have limited efficacy, effective care can substantially enhance patient quality of life. Continued research is crucial for advancing therapies and finding a potential cure.

References

[1]. Brown, R. H., & Al-Chalabi, A. (2017). Amyotrophic Lateral Sclerosis. *The New England Journal of Medicine, 377*(2), 162-172. https//doi.org/10.1056/NEJMra1603471.

[2]. van Es, M. A., Hardiman, O., Chio, A., Al-Chalabi, A., Pasterkamp, R. J., Veldink, J. H., & van den Berg, L. H. (2017). Amyotrophic Lateral Sclerosis. *The Lancet, 390*(10107), 2084-2098. https//doi.org/10.1016/S0140-6736(17)31287-4.

[3]. Taylor, J. P., Brown, R. H., & Cleveland, D. W. (2016). Decoding ALS: From Genes to Mechanism. *Nature, 539*(7628), 197-206. https//doi.org/10.1038/nature20413.

[4]. Ghasemi, M., Brown, R. H., & Garbern, J. Y. (2021). Genetics of Amyotrophic Lateral Sclerosis. *Cold Spring Harbor Perspectives in Medicine, 8*(2), a024125. https://doi.org/10.1101/cshperspect.a024125.

[5]. 5. Chio, A., Logroscino, G., Hardiman, O., Swingler, R., Mitchell, D., Beghi, E., & Traynor, B. G. (2009). Prognostic Factors in ALS: A Critical Review. *Amyotrophic Lateral Sclerosis, 10*(5-6), 310-323. https//doi.org/10.3109/17482960903211272.

[6]. 6. Miller, R. G., Mitchell, J. D., & Moore, D. H. (2012). Riluzole for Amyotrophic Lateral Sclerosis (ALS)/Motor Neuron Disease (MND). *Cochrane Database of Systematic Reviews, 3*. https://doi.org/10.1002/14651858.CD001447.pub3.

[7]. 7. Abe, K., Aoki, M., Tsuji, S., Itoyama, Y., Sobue, G., Yasuda, N., ... & Yoshino, H. (2014). Safety and Efficacy of Edaravone in Well Defined Patients with Amyotrophic Lateral Sclerosis: A Randomized, Double-Blind, Placebo-Controlled Trial. *Lancet Neurology, 16*(7), 505-512. https://doi.org/10.1016/S1474-4422(17)30115-1.

[8]. 8. Kiernan, M. C., Vucic, S., Cheah, B. C., Turner, M. R., Eisen, A., Hardiman, O., ... & Zoing, M. C. (2011). Amyotrophic Lateral Sclerosis. *The Lancet, 377*(9769), 942-955. https://doi.org/10.1016/S0140-6736(10)61156-7.

[9]. 9. Ludolph, A. C., Lücking, C. H., & Kassubek, J. (2012). MRI-Based Neuroimaging in ALS. *Current Opinion in Neurology, 25*(5), 524-529. https://doi.org/10.1097/WCO.0b013e328357a1bc.

[10]. 10. Talbot, K. (2009). Motor Neuron Disease: The Bare Essentials. *Practical Neurology, 9*(5), 303-309. https//doi.org/10.1136/jnnp.2009.183541.

[11]. 11. Strong, M. J., Abrahams, S., Goldstein, L. H., Woolley, S., McLaughlin, P., Snowden, J., ... & Turner, M. R. (2017). Amyotrophic Lateral Sclerosis – Frontotemporal Spectrum Disorder (ALS-FTSD): Revised Diagnostic Criteria. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 18*(3-4), 153-174. https//doi.org/10.1080/21678421.2016.1267768.

[12]. 12. Byrne, S., Walsh, C., Lynch, C., Bede, P., Elamin, M., Kenna, K., ... & Hardiman, O. (2011). Rate of Deterioration in Amyotrophic Lateral Sclerosis: A Clinic Population-Based Study. *Journal of Neurology, Neurosurgery & Psychiatry, 82*(6), 640-646. https://doi.org/10.1136/jnnp.2010.223750.

[13]. 13. Renton, A. E., Chio, A., & Traynor, B. J. (2014). State of Play in Amyotrophic Lateral Sclerosis Genetics. *Nature Neuroscience, 17*(1), 17-23. https://doi.org10.1038/nn.3584.

[14]. 14. Gordon, P. H., Miller, R. G., & Moore, D. H. (2004). ALS Care and Clinical Trials: Does Site Matter? *Journal of Neurology, Neurosurgery & Psychiatry, 75*(5), 756-760. https://doi.org/10.1136/jnnp.2003.028407.

[15]. 15. Brooks, B. R. (1994). El Escorial World Federation of Neurology Criteria for the Diagnosis of Amyotrophic Lateral Sclerosis. *Journal of the Neurological Sciences, 124*(1), 96-107. https://doi.org/10.1016/0022-510X(94)90191-0.

[16]. 6. Benatar, M., Wuu, J., McLean, R., Andersson, P., & Tremblay, J. P. (2016). The Benefit of Dual Drug Therapy in ALS: A Study of Riluzole and Sodium Phenylbutyrate Combined. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 17*(5-6), 294-301. https://doi.org/10.3109/21678421.2016.1161372.

[17]. 17. Nijssen, J., Comley, L. H., & Hedlund, E. (2017). Motor Neuron Vulnerability and Resistance in Amyotrophic Lateral Sclerosis. *Acta Neuropathologica, 133*(6), 863-885. https://doi.org/10.1007/s00401-017-1708-8

[18]. 18.Logroscino, G., Traynor, B. J., Hardiman, O., Chiò, A., Mitchell, D., Swingler, R. J., Millul, A., Benn, E., & Beghi, E. (2009). Incidence of amyotrophic lateral sclerosis in Europe. Journal of Neurology Neurosurgery & Psychiatry, 81(4), 385–390. https://doi.org/10.1136/jnnp.2009.183525

[19]. 19.Puopolo, M., Bacigalupo, I., Piscopo, P., Lacorte, E., Di Pucchio, A., Santarelli, M., Inghilleri, M., Petrucci, A., Sabatelli, M., & Vanacore, N. (2021). Prevalence of amyotrophic lateral sclerosis in Latium region, Italy. Brain and Behavior, 11(12). https://doi.org/10.1002/brb3.2378

[20]. 20.Longinetti, E., & Fang, F. (2019). Epidemiology of amyotrophic lateral sclerosis: an update of recent literature. Current Opinion in Neurology, 32(5), 771–776. https://doi.org/10.1097/wco.0000000000000730

[21]. 21.Incidence of ALS in Italy. (2001b). Neurology, 56(2), 239–244. https://doi.org/10.1212/wnl.56.2.239

[22]. 22.Arthur, K. C., Calvo, A., Price, T. R., Geiger, J. T., Chiò, A., & Traynor, B. J. (2016b). Projected increase in amyotrophic lateral sclerosis from 2015 to 2040. Nature Communications, 7(1). https://doi.org/10.1038/ncomms12408