# A Sporadic Case of Amyotropic Lateral Sclerosis Presenting As Pseudoblbar Palsy – A Case Study

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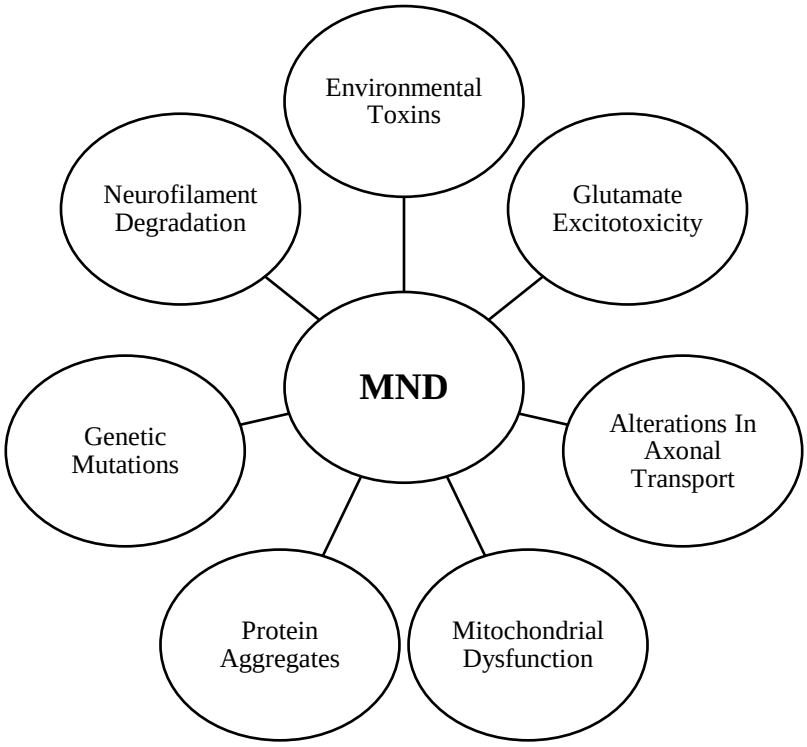
## Introduction:

Motor neuron diseases are progressive neuro-degenerative disorders of the motor neurons in the brain and spinal cord, that controls the voluntary movements of the muscles [1,2]. Signals are transmitted from the upper motor neurons (UMN) in the brain to the lower motor neurons (LMN) in the spinal cord and then to different muscles [2]. Motor neuron diseases can be hereditary or acquired and vary in its clinical presentation [7]. MND is quite rare with an incidence of almost 2/1,00,000 people in most parts of the world and the incidence is higher in males than the females [3,4,5]. Onset usually occurs at an age of 40 – 60 years [3].

Pathologic factors that contribute to MND are,




Clinically ALS presents with different symptoms based on the involvement of UMN or LMN. Upper motor neurons show signs as spasticity, hyperreflexia, Babinski sign, sparse wasting and lower motor neurons present as atrophy, flaccidity, hyporeflexia or areflexia, low threshold for irritation of motor neurons.

Although there is no cure for ALS, symptomatic and supportive care can be given for the patients. Riluzole is the only drug available from 1996 for the treatment of ALS. It works by reducing the glutamate excitotoxicity [1,6]by producing an inhibitory effect on glutamate release. Increase in the levels of glutamate in the synapse causes excitotoxicity. *Miller et al.*(2012) found that taking 100 mg of riluzole daily probably prolongs median survival by two to three months. Other treatment options available are gene therapy, stem cell therapy, growth factor therapies and various other supportive treatments are helpful.

## CASE REPORT:

A female patient of age 59 years presented with complaints of giddiness, weakness in hand grip, cough and difficulty in speech for the 6 months before diagnosing with ALS. Motor Neuron Conductivity (MNC), Sensory Neuron Conductivity (SNC) and electromyography (EMG) study favored the diagnosis of ALS. She was on Riluzole 50 mg 1-0-1. She developed progressive degeneration and had complaints of difficulty in using her left fingers and dysphagia confirming the involvement of bulbar phenotype. She had choking that caused aspiration pneumonia and transferred to ICU with breathing difficulty. Progressive motor weakness and bulbar dysfunction lead to premature death, frequently from respiratory failure [2] Impaired bulbar function cause aspiration and dyspnoea may be due to infection [7].

On examination, she was tachypnoeic, tachycardic, and ECG showed sinus tachycardia with short PR interval. Her blood investigations showed evidence of infection. She was found to have consolidation of right lung with progressive deterioration of sensorium and developed respiratory failure. Respiratory symptom is common in MND, that develops because of weakness in respiratory muscles. Tracheostomy was done and respiration was supported by a BiPAP machine with 15 liters of oxygen.

| CBC and COAGULATION PROFILE | |
|---|---|
| Haemoglobin | 11.8 g/dl |
| WBC | 24,520 cells/cu.mm |
| Polymorphs | 93.6% |
| Lymphocytes | 4.2% |
| Monocytes | 2.1% |
| PT time | 15 seconds |
| INR | 1.1 |
| APTT | 56 seconds |

| RENAL FUNCTION TESTS & ELECTROLYTES | |
|---|---|
| UREA | 40 mg/dl |
| CREATININE | 0.5 mg/dl |
| SODIUM | 157 mEq/l |
| POTASSIUM | 4.1 mEq/l |
| CHLORIDE | 114 mEq/l |
| BICARBONATE | 33 mEq/l |
| MAGNESIUM | 2.0 mEq/l |

| LIVER FUNCTION TESTS | |
|---|---|
| Total bilirubin | 1.4 mg% |
| Direct bilirubin | 1.0 mg% |
| Indirect bilirubin | 0.4 mg% |
| SGOT | 54 U/L |
| SGPT | 51 U/L |
| Total protein | 7 mg% |
| Albumin | 3.1 mg% |
| Globulin | 3.8 mg% |

Her vitals were elevated with a temperature of 101˚ F and pulse rate was 122 beats / min. ABG analysis had a blood pH of 7.35, $pCO_2$ -35.3, $pO_2$ -56, $HCo_3$ - 22.9, lactate - 1.1 on admission and $CO_2$ retention was more inside and value got increased gradually clearly indicating respiratory muscle paralysis.

Patient's kidney was gradually deteriorating with elevating urea levels and alterations in the electrolyte levels. She was given supportive treatment with antibiotics, bronchodilators and corticosteroids

**DISCUSSION:**

ALS is a progressive neurodegenerative disorder that affects the motor neurons and voluntary movements are lost gradually [1,3,7]. Patient has a life time of 3 to 5 years after diagnosing ALS. MND is a very rare presentation of about 2/1,00,000 cases in most parts of the world. Of these cases sporadic form of ALS (SALS) is common than familial ALS (FALS) [1]. Etiology of MND is not explained clearly, genetics, environmental and occupational risks offer a major contribution. Mutation in Superoxide dismutase 1(SOD1) is the major contributor for nerve damage and various other genes as Fused sarcoma (FUS), chromosome 9 open reading frame 72 (C9orf72), Transactive Response DNA binding protein (TARDBP) and 16 other gene mutations also contribute to MND. Patient diagnosed with ALS have limb onset weakness in about 80% of the cases and only 20% have bulbar onset [1]. Once the disease is diagnosed there are chances of poor prognosis when respiratory complications and bulbar phenotype. Patient will report weakness may not mostly recognize muscle wasting and fasciculations. Weakness may further spread to opposite limb or to another region of brain or spinal cord and may worsen the prognosis. Spasticity, weakness and hyperreflexia are seen in upper motor neuron damage and reduced tone, fasciculations, reduced muscle stretch are seen in lower motor neuron damage. Some patients may predominantly have upper motor signs while some have report lower motor signs. Combination of both upper and lower motor signs are seen in bulbar onset ALS [2]. Electromypgraphy (EMG), and nerve conduction studies for both sensory and motor neurons will propose the appropriate diagnosis of MND. Other investigations such as MRI brain and spinal cord, lumbar puncture, routine blood test and thyroid function tests should be done to rule out the mimics. Riluzole is the only FDA approved treatment option available for ALS. Other treatment options should be symptomatic and supportive. Multidisciplinary care provides better options to improve the quality of life.

**REFERENCES:**


1. Talbot K. Motor neuron disease. *Postgrad Med J.* 2002; 78: 513-519.
2. Tiryaki E, Horak HA. ALS and other motor neuron diseases. *Continuum (Minneap Minn)* 2014; 20 (5): 1185 – 1207.
3. Vucic S. The varied motor neuron disease phenotypes. ACNR; 2014: 14(4): 27–28.
4. Sathasivam S. Motor neuron disease: clinical features, diagnosis, diagnostic pitfalls and prognostic markers. *Singapore Med J.* 2010; 51(5): 367-372.
5. Boillee S, Velde VC, Cleveland WD. ALS: A Disease of Motor Neurons and Their Nonneuronal Neighbours. *J.neuron.* 2006; 52: 39-59.
6. Longo, Fauci, Kasper *et al.* Harrison's principles of internal medicine. 18th edition, McGraw-Hill chapter 37: 6579-6591.
7. Motor neuron diseases US department of health and human services. NIH publications No, 12-5371 March 2012.