DELAY IN DIAGNOSIS OF MOTOR NEURON DISEASE. A TERTIARY CARE EXPERIENCE

Hazim Brohi[1], Naveed Uddin Ahmed[2], Rajesh Khatri[3], Ishaq Asghar[4]
[1] Associate Professor Neurology Department LNH Karachi
[2] Assistant Professor Neurology Department LNH Karachi
[3] Resident Neurology Department LNH Karachi
[4] Intern LNH Karachi

Correspondence to: Neurology Department, Liaquat National Hospital, Karachi, Pakistan. Email address: hazimbrohi@yahoo.com.

ABSTRACT

Motor Neuron Disease is a progressive neurological disorder with poor prognosis. Inability to correctly recognize the disease, leads to undue agony and diagnostic dilemma. Studies have been done to look at the average duration taken to diagnose the disease from the onset of symptoms. The literature shows that latency from symptom onset to diagnosis range from 10.6–17.5 months. No such study has been conducted in our region. We reviewed 62 patients referred to the neurophysiology lab from year 2009 to 2013 with quarry of MND. Only 40 patients were identified to fulfill the El Escorial criteria for MND. The median time of diagnosis from onset of symptoms was one year. This is almost similar to that of world literature. We conclude that in spite of paucity of heath service and high rate of illiteracy the timeliness of diagnosis was consistent with the rest of the world.

INTRODUCTION

Motor Neuron Disease (MND) is a progressive neurodegenerative disease that usually occurs in the fifth to sixth decades. The duration of survival is short, with an average life expectancy of less than 3 years after diagnosis[1]. Recent few studies have shown that the incidence and mortality rates of ALS have increased over the past several decades, possibly owing to ageing of the population[2-3]. Timely diagnosis is an essential component of high quality health care; particularly confirm a diagnosis as devastating as MND. Early diagnosis of MND is very important as significant psychological stress accompanies the period awaiting a diagnosis[4]. A study by Johnston et al reported that the majority of MND patients described positive attitude when 'labelled' for their condition and that the earlier diagnosis allows these patients to have more time to make personal and financial adjustments and make plans for the future, including modifications in their homes to cope with impending disability and co formulating working plan[5]. The average latency from symptom onset to diagnosis in MND is about 14 months, about one third of expected survival. Occasionally, survival following diagnosis may be less than six months so a diagnosis is usually required before care can be organized[6]. According to the data from Ireland Motor Neuron disease the duration of diagnosis of MND from the onset was 15.6 months[4]. Our literature search revealed no such study from our region. No details are available regarding time taken for the diagnosis of this devastating disease.

METHODS

As NCS and EMG is the diagnostic test in spite of clinical suspicion, we assumed that the final label can only be given once the test has been done. So the time between the onset of symptoms and diagnosis will be" the time the patient becomes significantly symptomatic to seek medical help to the time he/she is labelled as suffering from MND and that's when his/her electrophysiological study is done. This was a retrospective study. We reviewed the charts of all patients who came to neurophysiology laboratory at Liquat National Hospital with suspicion of MND from year 2009 to year 2013. The charts used for recording the data were reviewed for details of the patient like onset of symptoms, age, gender and the EMG details. The data was then entered on a self designed questionnaire. Patients fulfilling EL Escorial criteria were considered as suffering from MND. SPSS version 18 was used for analysis of the results.

RESULTS

Total of 62 patients were referred to rule out motor neuron disease. Eight patients did not show anterior horn cell

involvement. Out of remaining Fifty four patients 40 patients i.e. 64% fulfilled the El Escorial criteria for MND, 13 patients i.e. 21% had suggestion of anterior horn cell involvement but did not have 3 areas involved as per El Escorial criteria. The median time of diagnosis from onset of symptoms was 1 year. 67 % patients had tongue involvement and 82 % had spine involvement at time of diagnosis. Limb involvement was almost 95%.

DISCUSSION

Our literature search failed to show any study regarding time lapsed between onset of symptoms and diagnosis of MND from our region. As most patients with ALS die of respiratory failure, usually within 3 to 5 years from the onset of symptoms, the time is limited for patient. Even in well developed countries the time to diagnosis is prolonged. According to a study from Ireland the survival time from diagnosis to death was 16.4 months (5) and the diagnostic time was 15.6 months. As no such information was available from third world countries, the diagnostic delay may be very long. We conducted this study to see the time to diagnosis in our setup. Surprisingly the median time of diagnosis was one year. A detailed review of the data showed a diverse range of time ranging from 1 month to 4 years. We believe the short duration from onset may not be true representative as certain patients are illiterate and may not have noticed the disease until late or due to lack of proper diagnosis, the patient may have been roaming around till it was too late. However the median time was one year, which seems to be in accordance with literature quoted from the western countries. Another important observation was the fact that around 21 % patients were not labelled as motor neuron disease as only El Escorial criteria was used for the diagnostic purposes. Recently Awagi Shiama criteria is being used which is less stringent and more patients may have qualified for diagnosis of MND using these criteria7. The median time, when including all patients with anterior horn cell was 9 months. According to the study conducted in Ireland, which is a community based study, the correct diagnosis was made in 95 % patients by neurologist compared to 5 % patients seen by general physicians4. Thus properly referred patients will have less diagnostic delay. We conclude that greater public awareness especially among health care professionals such as primary care physicians and readily availability of neurologists may save patient from the agony of diagnostic delay.

Chart 1: Numbers of patients fulfilling El Escorial criteria

	Number of patients	Percent
Patient fulfilling criteria	40	64.5
Not fulfilling Criteria	13	21.0
Patient with other Diagnosis	9	14.5
Total number of patients	62	100

Chart 2: Median time from onset to diagnosis

Fulfill El Escorial Criteria	1 Year
All Anterior horn cell	9 Months

Chart 3: Details of time duration

Duration	No of Patients	Percent
1 Months	1	2.5
2 Months	3	7.5
3 Months	5	12.5
4 Months	1	2.5
5 Months	3	7.5
6 Months	3	7.5
8 Mont hs	1	2.5
9 Months	2	5.0
1 Year	6	15.0
1.5 Year	3	7.5
2 Years	2	5.0
2.5 Years	1	2.5
3 Years	3	7.5
4 Years	1	2.5
Not mention	5	12.5
Total	40	100

Table 4: Details of EMG findings (denervation, high broad MUAP)

Site involvement	Frequency	Percent
tongue	27	67.5
spine	33	82.5
Right upper limb	49	90.7
Left upper limb	45	83
Right lower limb	47	87
Left lower limb	47	87

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