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General Medicine

A RARE CASE OF MADRAS VARIANT OF MOTOR NEURON DISEASE

Dr Shamli Mishra 3rd Year Resident, Department Of General Medicine, VIMSAR, Burla.

Prof Dr. Manoj Kumar Mohapatra* Professor, Department Of General Medicine, VIMSAR, Burla. *Corresponding Author

ABSTRACT **INTRODUCTION:** Madras variant MND is a rare entity is characterized by multiple lower cranial nerve palsies(7-12) and atrophy of limbs. **BACKGROUND:** A 26 year old male presented with complaints of difficulty in hearing and hoarseness of voice since 1 year and weakness of lower limbs since 10 days without any sensory, bowel and bladder symptoms. On examination, his vitals were stable. Cranial nerves examination revealed abnormality in the form of b/l sensorineural hearing loss, weakness of b/l facial muscles with fasiculations, absent gag reflex with atrophy and fasiculations of the tongue. There was significant wasting of muscles of face, upper and lower limbs with hypotonia in upper and hypertonia in lower limbs. Reflexes were feeble in upper limb and exaggerated in lower limb. On investigation EMG-NCS showed chronic partial denervation of all muscles with reduced recruitment. **CONCLUSION:** Very few cases of Madras variant of MND have been reported due to rare combination of features and often misdiagnosed.

KEYWORDS :

INTRODUCTION:

Madras variant of motor neuron disease in a rare variant of MND which was first identified in the year 1970 and was named by Meenakshisundaram et al to a group of patients from Madras. This disease is characterized by typical constellation of symptoms i.e. younger age of onset, weakness and atrophy especially of the distal muscles in the limbs, multiple lower cranial nerve palsy, pyramidal dysfunction and sensorineural hearing loss along with optic atrophy rarely.

CASE SUMMARY:

A 26 year old male, youngest of all siblings, born of non consanguineous marriage presented with the chief complaints of weakness of both halves of the face since 2 years, hearing loss since 1 year, difficulty in protrusion of tongue, swallowing of liquids and hoarseness of voice since 6 months and difficulty in walking since 1 month with no sensory ,bowel or bladder abnormalities , no significant past history. Family history suggests similar illness in the older sibling who died at the age of 25 years despite treatment.

On examination – Patient was thin built, conscious and cooperative, with normal vitals. Higher mental functions were normal except for his speech which was flaccid dysarthric type. Cranial nerve examination revealed abnormality in the optic nerve where in the visual acuity was 6/36 and fundoscopy revealed b/l optic atrophy. Facial nerve examination revealed weakness of muscles of facial expressions with atrophy and fasciculations, loss of nasolabial folds and positive bells phenomena. 8th cranial nerve examination suggested positive Rinnes with b/l sensorineural hearing loss in PTA. There was absence of gag reflex with hoarseness of voice on examination of 9th and 10th CNs. The tongue showed diffuse atrophy and fasciculations.

Motor system revealed power of grade 4 in distal muscles of upper limbs, grade 3 in the distal muscles of lower limbs with atrophy and fasciculations. DTRs had a grade of 1+ in biceps, triceps with absent supinator, 3+ in knee and ankle with b/l plantar extensor. Sensory system, coordination, cerebellar signs, spine and other systems were normal.

INVESTIGATIONS:

CBC/LFT/RFT/ RBS – Normal. HIV, HBV,HCV- Non reactive. Vitamin B12 assay – 1500pg(Normal). CXR, USG abdomen- normal. MRI of cervical spine- Mild cervical spondylosis without compression of nerve roots or spinal canal stenosis.

NCS- normal. EMG- chronic denervation in facial muscles, distal upper and lower limb. The calf muscles showed fibrillation and persistent fasciculations.

DISCUSSION:

Madras MND is a rare entity and so far only 200 cases have been reported with an average incidence of <1/1000000, mostly from southern India. The interesting aspect of this case is that this has been reported from eastern India , where the incidence is minimal. This patient has features of early and severe involvement of facial muscles in addition to bulbar involvement which distinguishes him bulbar variant of ALS. Optic atrophy is present in 20% of cases and b/l SNHL in 91% cases, both of which are present in the patient.

Patient is now on followup since the last 8 months with oral Riluzole therapy and the rate of progression has been quite slow as compared to other ALS patients.

REFERENCES

1. Gourie-Devi, M., & Nalini, A. (2003). Madras motor neuron disease variant, clinical features of seven patients. Journal of the Neurological Sciences, 209(1-2), 13–17.
2. Gourie-Devi, M., Maheshwari, S., Panda, A., & Bala, K. (2013). Chin fasciculations in Madras motor neuron disease: A new clinical feature. Neurology India, 61(6), 653.
3. Meenakshisundaram E, Jagannathan K, Ramamurthi B. Clinical pattern of motor neuron disease seen in younger age groups in Madras. Neurol India. 1970;18(Suppl.1):109-112.
4. Valmikinathan K, Mascreen M, Meenakshisundaram E, Snehalatha C. Biochemical aspects of motor neurone disease--Madras pattern. J Neurol Neurosurg Psychiatry. 1973;36:753-756.
5. Shankar SK, Gourie-Devi M, Shankar L, Yasha TC, Santosh V, Das S. Pathology of Madras type of motor neuron disease (MMND)--a histological and immunohistochemical study. Acta Neuropathol. 2000;99:428-434.
6. Nalini A, Yamini BK, Gayatri N, Thennarasu K, Gope R. Familial Madras motor neuron disease (FMMND): study of 15 families from southern India. J Neurol Sci. 2006;250:140-146.
7. Nalini A, Thennarasu K, Yamini BK, Shivashankar D, Krishna N. Madras motor neuron disease (MMND): clinical description and survival pattern of 116 patients from Southern India seen over 36 years (1971-2007). J Neurol Sci. 2008;269:65-73.