

Madras Variant Motor Neuron Disease - A Rare Presentation



Medical Science

KEYWORDS : Madras variant, sensorineural deafness, optic atrophy.

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ABSTRACT

Madras variant MND is a rare variant of Madras MND. It is characterised by atrophy and weakness of limbs, multiple cranial nerve palsies particularly 7,9 to 12, sensorineural hearing loss, optic atrophy. We present a case of madras variant of Madras MND who had typical constellation of paralysis of upper and lower motor neurons with combination of sensorineural deafness and optic atrophy.

INTRODUCTION:

Motor neuron disorders are a group of neurodegenerative disorders and arguably the most devastating group of nervous disorders. They are characterised by death of lower motor neurons and upper or corticospinal motor neurons⁴. The death of neurons in brainstem and spinal cord leads to denervation and consequent atrophy of muscle fibres. The end stage of these disorders is marked by respiratory muscle paralysis and prognosis is uniformly poor in all types of motor neuron disorder.

CASE REPORT:

40 year old male presented to us with chief complaints of hearing loss since 8 years, difficulty in walking since 2 years, dysarthric speech since 4 months. About 8 years ago he started to notice decreased sense of hearing, initially for low tones but later even for higher tones. Later he even failed to decipher what others spoke. About 2 years back he started having difficulty in walking, and experienced tingling sensation in both lower limbs. Later he experienced clumsiness of feet while walking, slippage of footwear with his knowledge. He had difficulty in climbing stairs, frequent falls from stubbing of toes during walking, he could not button/unbutton his shirt, later he could not stand from sitting position, difficulty in holding objects. About 2 months back he started experiencing difficulty in speaking in form of dysarthria. Patient also had impaired vision in right eye since about 15 years. Family history- Father had similar gait abnormalities, sister has gait abnormalities and hearing impairment. Son had similar gait abnormalities. No any other systemic complaints were present. No h/o of high grade fever, headache and vomiting, convulsions, diplopia, ear discharge. No past history of trauma. No past h/o of hypertension, diabetes, ischemic heart diseases, stroke.

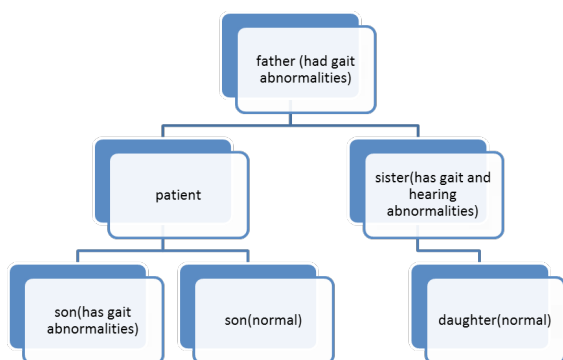


Fig 1: Pedigree chart of patient.

On examination ; Temperature was normal, Pulse was 74/min, BP-116/70mm hg, Respiratory/CVS/Per abdomen-normal. CNS; Patient was conscious, alert, co-operative. Higher mental revealed a right handed person with dysarthric speech, oriented to time place and person. Memory tests were normal. Cranium and Spine appear normal. No signs of neck rigidity. Cranial nerves examination was normal except for presence of bilateral sensorineural hearing loss which was confirmed on pure tone audiometry. Fasciculations were present on tongue. Motor System revealed Power of grade 4 in bilateral upper limbs, grade 3 in bilateral proximal muscles of lower limbs, grade 2 in distal muscles of lower limbs, mild atrophy of forearm and calf muscles, wasting of muscles over dorsum of right hand and wasting over dorsum of bilateral foot muscles, hammer toes of bilateral foot, co-ordination preserved, tone was normal in all limbs. Sensory System; superficial, deep, cortical sensations were normal.

Reflexes:

	Biceps	Triceps	Supinator	Knee	Ankle
Right	1+	1+	absent	absent	absent
Left	1+	1+	Absent	absent	Absent

Plantar; Bilateral Extensor. There were no bowel and bladder movement abnormalities or trophic changes. There were no any cerebellar signs. A broad based gait was present. Investigations: CBC/ESR/RFT/LFT/RBS-Normal. Vitamin B12-346(normal). HIV/HbsAg-Nonreactive. Chest x ray/USG of Abdomen, KUB, Prostate-normal. Audiogram-bilateral moderate to severe sensorineural hearing loss. EMG-NCV-evidence of chronic partial denervation in all muscles of both upper limbs and both lower limbs examined with reduced recruitment pattern. Fasciculation potential are seen in all these muscles. There is evidence of denervation in tongue.

Fundus:

Right Eye	Left EYE
Pale disc	Small disc
Choroidal coloboma	Brown cataract
RAPD	

findings are suggestive of optic atrophy

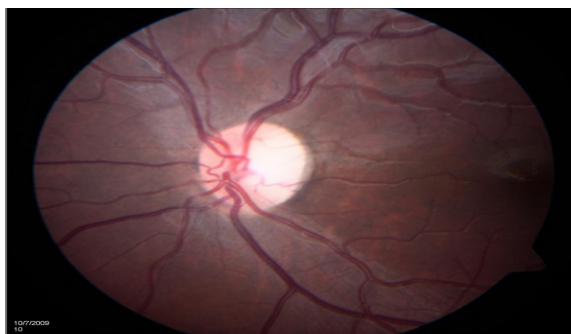


Fig 2:Suggestive of optic atrophy

MRI Brain and LS Spine was normal. Based upon clinical and laboratory findings we made a provisional diagnosis of Madras variant of MND. We started the patient on Tab Riluzole, multivitamin supplements, physiotherapy was advised.

Discussion

Madras MND was originally identified in 1970 in southern city of Madras¹. Until 2010, 152 cases have been reported. Outside this core region solitary cases are reported in China, Italy, Korea and Thailand. Pattern of inheritance in familial Madras MND appears to be autosomal recessive, with few autosomal dominant, reported in Indian literature³.

Types: 1) Sporadic MMND. 2) Sporadic MMND variant. 3) Familial MMND. 4) Familial MMND variant.

Salient features of Madras MND—Younger age of onset, wasting, weakness mainly of distal muscles, bulbar dysfunction and facial muscle involvement, pyramidal dysfunction, sensorineural hearing impairment, optic atrophy. Evidence of sensorineural impairment, optic atrophy are important features of MMND variant. Optic atrophy is documented in 20% of Madras MND patients and sensorineural deafness in 91% of patients², both features were present in our patient. But we could not get the exact figures about combination of both deafness and optic atrophy in Madras variant of Madras MND.

Pathogenesis: Low plasma citrate and elevated pyruvate levels resulting in low citrate to pyruvate ratio have been observed in some patients of MMND.

Treatment: No specific treatment available. IVIG and Riluzole have been tried with variable success.

CONCLUSION:

Madras variant of MND is a very rare variant and it requires high degree of suspicion and close follow up. Very few cases have been reported in recent years. The case report has been presented due to rare combination of features of motor neuron disease and sensorineural deafness with optic atrophy.

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