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A Case Report – MELAS Syndrome

Dr Hariom Meena*, Dr Ramraj Meena*, Dr Surendra Khosya*

Department of Medicine
Govt Medical College & Attached, MBS. Hospital, Kota, Rajasthan, India
*JR 3 IN DEPARTMENT OF INTERNAL MEDICINE
Email: drkhosya3@gmail.com

I. INTRODUCTION

MELAS (mitochondrial encephalomyopathy, lactic acidosis, and recurrent stroke-like episodes) syndrome is a rare neurodegenerative and fatal disease cause by mutation in mitochondrial DNA. Patients with mitochondrial myopathies typically have exercise- induced symptoms. Thus, patients are often advised to avoid exercise, which leads to deconditioning. Currently, the concept of aerobic exercise training as therapy for mitochondrial disease is not well established.

MELAS is a condition that affects many of the body's systems, particularly the brain and nervous system (encephalo-) and muscles (myopathy). In most cases, the signs and symptoms of this disorder appear in childhood following a period of normal development[3].Early symptoms may include muscle weakness and pain, recurrent headaches, loss of appetite, vomiting, and seizures. Most affected individuals experience stroke-like episodes beginning before age 40. These episodes often involve temporary muscle weakness on one side of the body (hemiparesis), altered consciousness, vision abnormalities, seizures, and severe headaches resembling migraines. Repeated stroke-like episodes can progressively damage the brain, leading to vision loss, problems with movement, and a loss of intellectual function (dementia).

Most people with MELAS have a buildup of lactic acid in their bodies, a condition called lactic acidosis. Increased acidity in the blood can lead to vomiting, abdominal pain, extreme tiredness (fatigue), muscle weakness, loss of bowel control, and difficulty breathing. Less commonly, people with MELAS may experience involuntary muscle spasms (myoclonus), impaired muscle coordination (ataxia), hearing loss, heart and kidney problems, diabetes, Epilepsy, and hormonal imbalances.
MELAS (mitochondrial encephalomyopathy, lactic acidosis, and recurrent stroke-like episodes) syndrome is a mitochondrial disorder. Patients with MELAS generally have a poor prognosis and outcome, as effective therapiesfor MELAS syndrome have not been established. The clinical management of individuals is largely supportive and includes exercise training.[1] In recent studies, exercise has been shown to be of possible benefit to patients with mitochondrial disease as it prevents deconditioning, which can exacerbate pre-existing exercise intolerance and fatigability[2,3].Aerobic training enhances aerobic capacity, muscle oxidative metabolism, and ATP production, decreases lactate levels, and improves quality of life in patients with mitochondrial disease.[4] A regular low intensity regimen can improve muscle endurance and cardiopulmonary function.

II. CASE REPORT

The patient, Ahsaan Mohd. 40-year-old man, was admitted to our hospital due to an episode involving visual hallucinations, migraine-like headaches, seizure, vomiting, recurrent sudden loss of consciousness, and relative lower limbs weakness.None of his family members or relatives had MELAS. His speech was fluent with normal comprehension and repetition. He had grade 4 muscle power of his both upper and lower limb, bilateral plantar are extensor. He had intact, brisk symmetrical deep tendon reflexes and absent Babinski sign bilaterally. On day 2 of his hospitalization, he become conscious and his altered sensorium improved and headache, vomiting completely resolved. On day 3 he walk a maximal distance of 5 meters. He had a wide-based, slightly ataxic gait as well.

Laboratory data included a serum lactate of 62.9 mmol/L (normal < 2.1 mmol/L). widal test and MP by card are normal. CSF done which is within normal limit. Serum phorphobilinogen level was normal. Magnetic resonance imaging (MRI) brain studies showed area of hyperintencity in periventricular ,parieto-occipital white matter s/o leukoencephalopathy .(Figure 1). Nerve conduction velocities (NCV) of bothlower limbs were normal, but, on electromyography (EMG), myopathic changes were seen in both lowerlimbs.. A muscle biopsy was taken from the rectus femoris muscle and ragged red fibers were noted on examination.

In the10 th day the patient's showed neurological improvement in muscle power, balance, endurance, and his gait speed. Her both extremity muscle power improved from grade 4 to grade 5. After discharge, the rehabilitation program prescribed for the patient emphasized instrumentaldaily activity and home aerobic exercise walked in his community 15-20 min daily. he coulddo household tasks, including housecleaning,bathing washing dishes, and changing bed linens with minimal difficulty. After one month he was readmitted in hospital with similar kind of illness.and improved within two days.

III. DISCUSSION

Figure 1. Magnetic resonance imaging study of the brain shows hyperintense T2 lesions predominantly in the subcortical white matter in the right temporal, parietal, and occipital lobes. Muscle strength, gait speed, physical function, and quality of life. The recovery was possible due to the muscle adaptation that occurs with exercise training, which may improve aerobic capacity, muscle oxidative metabolism, the mitochondrial

respiratory chain, and decrease secondary physical deconditioning.[5,6] The patient's ataxic gait pattern resolved, and he was able to walk with a normal gait due to improved muscle strength and decreased muscle fatigability. The patient progressed greatly in his daily functional skills. After discharge, the patient was prescribed a low level exercise program as part of her outpatient phase. The goal of our daily activity training is to minimize dependence and give the patient the ability to perform daily skill tasks. We evaluated the needs of the treatment phase based on the patient's physical status and provided an effective rehabilitation program.

Home aerobic exercise with minimal resistance can prevent fatigue and cardiopulmonary dysfunction. All of these results have been reported to be related to improvement of skeletal muscle oxidative capacity by extraction of available oxygen and enhancement of oxygen utilization by muscle[7].The treatment of mitochondrial disease has traditionally involved diets or drugs, such as coenzyme Q,cytochrome C, nicotinamide, dichloroacetate, and succinate; however, their effects are controversial. Rehabilitation in the form of aerobic training is another approach to the treatment of mitochondrial disease. It is thought that the capacity for oxygen extraction is directly correlated with the degree of impaired muscle O2 extraction, and that capillary density and mitochondrial oxidative capacity increase in skeletal muscle if adequate training is given.[6] The reported benefits of endurance training in patients with mitochondrial myopathy include notable improvement in work and oxidative capacity. Thus, muscle mitochondrial adaptations can improve the systemic a-vO2 difference, increase the extraction of oxygen during exercise, and may also allow patients to tolerate submaximal exercise.[6] Muscle strength, gait speed, physical function, and quality of life. The recovery was possible due to the muscle adaptation that occurs with exercise training, which may improve aerobic capacity, muscle oxidative metabolism, the mitochondrial respiratory chain, and decrease secondary physical deconditioning.[5,6] th and decreased muscle fatigability. Long term follow up of MELAS patients is required.

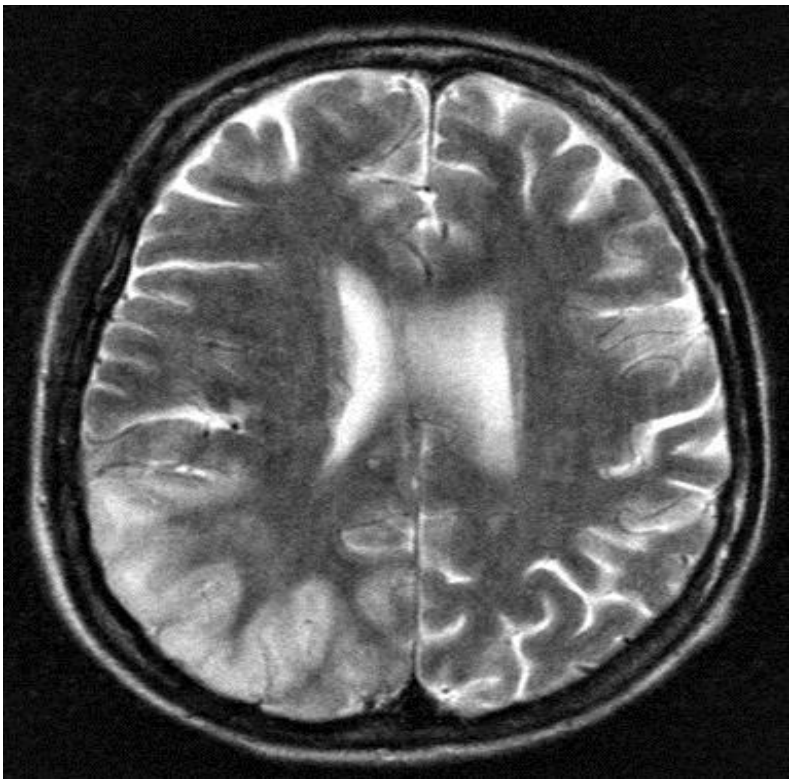

Figure 1

IV. CONCLUSION

Multidisciplinary management of pt. with MELAS required and would involve a geneticist, neurologist,cardiologist, nephrologist, ophthalmologist,Endocrinologist and dentist.

There is no known treatment of underlying disease which is progressive and fatal. Patients are managed according to what is affected at particular time. Metabolic therapies have been used to increase production of ATP. CoenzymeQ10 (CoQ10) ,ascorbate,riboflavin, vit k1 and vit k3, L-carnitin,succinate have proven quite successful.

Both pt. and family members should receive genetics counseling and the family should be educated about further deterioration and possible complication.

Clinical course is usually unpredictable with fluctuation and gradual determination leading to coma or death(usually from respiratory failure).

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