Mitochondrial Myopathy with Respiratory Dysfunction : A Case Report

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Abstract

A 50-year-old woman who presented with infective bronchiectasis and respiratory insufficiency was reported. Mitochondrial myopathy was diagnosed from a long history of chronic bilateral ptosis and external ophthalmoplegia with muscle wasting. Muscle biopsy revealed ragged-red fibers. After treatment with appropriate antibiotic and respiratory assisstance, the patient improved and returned to her usual state of health.

Key word : Mitochondrial Myopathy, Respiratory Dysfunction

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Mitochondrial disease can affect any organ system but usually involves skeletal muscle and brain. Ragged-red fibers in muscle biopsy specimens are an important marker for this disease. Chronic progressive external ophthalmoplegia (CPEO), characterized by chronic and progressive ptosis and limitation of eyeball movements, is a common manifestation but ventilatory failure is rarely the presenting manifestation[1-4]. We report a patient with mitochondrial myopathy who presented with predominant respiratory insufficiency.

CASE REPORT

A 50-year-old woman was admitted to Srinagarind Hospital in April 1999 with the symptoms of respiratory insufficiency. About 9 days earlier, she developed symptoms of fever, productive cough and dyspnea. She was admitted to a provincial hospital with a diagnosis of infective bronchiectasis and respiratory failure. She was intubated and ventilatory assistance was applied. After treatment with sulperazone to cover *Acinetobactor* infection, respiratory stasus improved. She

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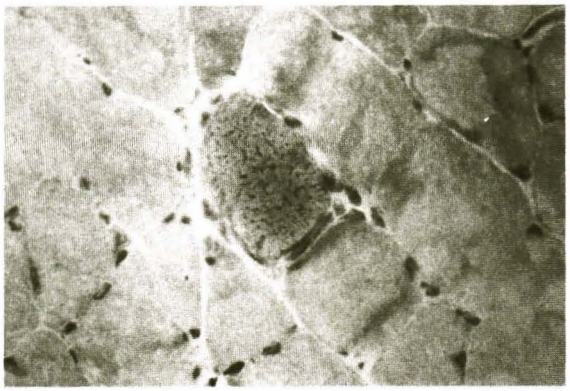

Fig. 1. Muscle biopsy with modified Gomeri trichrome stain reveals ragged-red fiber.

was transferred to our hospital for further investigation of wasting syndrome.

Relevant medical history revealed that she had a 30-year history of bilateral ptosis and an 8- year history of ophthalmoplegia and mild dysarthria. These symptoms had not fluctuated. Neurological examination revealed bilateral ptosis and external ophthalmoplegia, facial diplegia, mild proximal weakness, generalized hyporeflexia and generalized muscle wasting. Findings from the fundoscopic and other neurological examinations were normal. No known neurological disease was observed in her family. Prostigmine test was negative. Laboratory tests included, BUN, creatinine, electrolytes, calcium, phosphate, liver function tests, muscle enzymes, thyroid function tests, cerebrospinal fluid analysis and electrocardiogram were within normal limits. Electrodiagnostic study was normal for nerve conduction velocity and repetitive nerve stimulation. Needle electromyogram revealed signs of membrane instability and sign of early recruitment motor unit action potential (MUAP), which were suggestive of myopathy. Fresh frozen muscle biopsy of the left thigh with modified Gomeri trichome stain showed ragged-red fibers (Fig. 1). Electron microscopy revealed that there were increased numbers of mitochondria with disorganized myofibrils in the affected fibers. These mitochondrias were enlarged with aberration in shape and disordered cristae arrangement. Paracrystalline inclusion was not seen.

The patient was continuously treated with sulperazone. Her respiratory function improved gradually and artificial ventilation was discontinued at the 4th week of admission. On follow-up 4 months later, she had no symptoms of respiratory distress and had returned to her usual state of health.

DISCUSSION

In our case, the important clinical point that lead to the diagnosis of mitochondrial disease was a long history of chronic bilateral ptosis and external ophthalmoplegia (CPEO) with muscle wasting. Then muscle biopsy with modified Gomeri trichrome stain was performed to demonstrate ragged-red fibers. On the other hand, certain patients with mytochondrial myopathy and respiratory distress do not present with CPEO[3,4]. Also certain diseases, such as myasthenia gravis, thyroid disorders and muscular dystrophy can present with CPEO which should be primarily excluded.

The physiologic mechanisms of respiratory failure in this disease have been postulated. These are reduced ventilatory drive occurring especially in relation to surgery, sedation or intercurrent infection[2]; as in our case, and weakness and fatigue of the respiratory muscle[1].

In conclusion, we demonstrated a case of mitochondrial myopathy, presenting with respiratory distress, with a good clinical outcome. Therefore, mitochondrial myopathy should be ruled out in respiratory failure due to neuromuscular disorders.

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โรคกล้ามเนื้อผิดปกติชนิดไมโตคอนเดรีย ร่วมกับระบบการหายใจทำงานผิดปกติ : รายงานผู้ป่วย

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รายงานผู้ป่วยหญิงอายุ 50 ปี มาโรงพยาบาลด้วยโรคหลอดลมโป่งพองติดเชื้อร่วมกับมีระบบการหายใจล้มเหลว การวินิจฉัยโรคกล้ามเนื้อผิดปกติชนิดไมโตคอนเดรีย จากการที่มีประวัติของหนังตาตกและตากลอกไม่ได้ทั้งสองข้าง และกล้ามเนื้ออ่อนมาเป็นเวลานาน การตรวจทางจุลพยาธิของกล้ามเนื้อพบลักษณะ ragged-red fiber ผู้ป่วยมีอาการดีขึ้นเป็นปกติหลังการรักษา

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