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Mitochondrial Myopathy Presenting with Recurrent Lactic Acidosis and Diabetes Mellitus: A Diagnostic Challenge

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Abstract

Mitochondrial myopathies are rare disorders of oxidative phosphorylation that may manifest with muscle weakness, lactic acidosis, and multi-organ involvement. Endocrine dysfunction, particularly diabetes mellitus, is a recognized association.

We report a 28-year-old male with recurrent episodes of vomiting, myalgia, and altered sensorium, each associated with severe lactic acidosis. He had been diagnosed with diabetes mellitus at age 23, with poor glycemic control despite therapy. Examination revealed bilateral ptosis, proximal muscle weakness, and fatigability. Investigations showed persistent hyperlactatemia, elevated creatine kinase, and basal ganglia calcification. Muscle biopsy could not be performed locally, and the patient was referred to the National Institute of Neurosciences & Hospital, Dhaka, for further evaluation. He was managed with insulin, supportive care, avoidance of metformin, and physiotherapy.

This case highlights the diagnostic challenge when mitochondrial myopathy presents with endocrine manifestations such as diabetes. Awareness is crucial to avoid mismanagement and delayed recognition, particularly in resource-limited settings. *[J Assoc Clin Endocrinol Diabetol Bangladesh, 2025;4(Suppl 1): S53]*

Keywords: Mitochondrial myopathy, Lactic acidosis, Diabetes mellitus, Metabolic disorder

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