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General Medicine

METABOLIC ACIDOSIS:AN INTERESTING CASE PRESENTATION

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ABSTRACT Metabolic acidosis is a disorder frequently encountered in emergency medicine and intensive care medicine.Here we present a patient with repeated metabolic crisis, receiving several treatment in the past and not responding to medications.It was a diagnostic challenge. A middle aged male patient who was diagnosed with madelung's disease in the past, came to our hospital with exertional breathlessness, muscle fatigue and metabolic acidosis. Patient had previous admissions for similar symptoms. His investigations revealed high LDH and pyruvate levels with persistent lactic acidosis. A diagnosis of mitochondrial myopathy was confirmed with muscle biopsy. Patient showed gradual improvement with coenzyme Q10,vitamin E,omega 3 fatty acid,carntine and folic acid. This case demonstrates that patient with recurrent metabolic acidosis may have symptoms related to mitochondrial myopathy.

KEYWORDS : metabolic acidosis,lactic acidosis,mitochondrial myopathy,madelung's disease

INTRODUCTION

Metabolic acidosis is defined by the presence of acid base imbalance characterized by the presence of bicarbonate level less than 20 mmol/l. It is often described severe when ph is less than 7.2.

Acute metabolic acidosis is associated with organ failure,in particular respiratory and cardiovascular(1). Patients with elevated anion gap metabolic acidosis have elevated lactate levels,seen in severly ill patients and can be used to guage response to therapeutic interventions. Normally the principal sources of this acid are erythrocytes(which lack enzyme for aerobic oxidation),skeletal muscle,skin and brain.Hyperlactatemia occurs when lactate production exceeds lactate consumption. The etiologies of lactic acidosis is divided into type A(related to tissue hypoperfusion) and type B. Type B is divided into type B1(underlying primary diseases),type B2(medications and toxins) and type B3(inborn errors of metabolism).The main clinical features of lactic acidosis is marked hyperventilation. Aggressive treatment of the precipitating cause of lactic acidosis is the main component of therapy.The prognosis in most cases is that of the primary disorder that produced the elevated lactate levels(2,3).

We present the case of a patient in whom recurrent episodes of metabolic acidosis were related to an underlying mitochondrial disorder. Treatment of the mitochondrial disorder resulted in resolution of symptoms.

Case Presentation

This patient presented to our facility with a fairly long eventful medical history.initially , patient was diagnosed with madelung's syndrome(multiple symmetric lipomatosis). Subsequently,he developed fatigue and progressive weakness,the cause of which remain unclear. He had an admission for suspected acute coronary syndrome,but coronary angiogram was normal.he later developed a second episode of metabolic acidosis and generalized weakness, a diagnosis of guillian barre syndrome was made.he was evaluated in our facility in view of exercise intolerance,fatigue and proximal muscle weakness since 1 month. Clinical examination revealed grade 3 power in proximal muscles of upper limb and lower limb,the rest of the CNS was normal.routine blood and urine investigations were normal. Serum CPK,lactate and Ldh levels were high.serum lactate pyruvate ratio was high. Muscle biopsy showed ragged red fibres,ragged blue fibres and few COX deficient fibres suggestive of mitochondrial myopathy. Our patient was started on a compound of coenzyme Q10 200 mg,creatine 1000 mg,carnitine 200 mg and folic acid 1 mg.he gradually showed significant improvement in his symptoms over a course of few months.

DISCUSSION

Mitochondrial myopathies are disorders characterized by morphological abnormalities of muscle mitochondria. Mitochondrial diseases can affect multiple organ systems.Involvement of brain,skeletal system,peripheral nervous system,eyes and gastrointestinal system are characteristic. Although earlier considered to be a rare class of disorders,the recent epidemiological studies suggest that 1 in 5000 individuals are being affected by mitochondrial dysfunction and diseases[5]. Though the prevalence of individual mutations is much higher, approaching 1 in 200 live births[6], only a small proportion of individuals harbouring these mutations develop disease. In addition, features of multiple lipomatosis(madelung's disease)and short stature may contribute in making the clinical diagnosis. Elevated lactate levels,metabolic crisis with minor stresses and impaired glucose tolerance are helpful laboratory features(7). In most cases , a constellation of these various features help the clinician to suspect this uncommon diagnosis. The disorder in an an individual patient may fit into a well defined mitochondrial syndrome like MELAS or maybe non syndromic as in our case. Making a diagnosis of mitochondrial disorder is important in many ways. As in our case,the varied presentation may easily be mistaken for another disorder. An accurate diagnosis avoids unnecessary investigations and potentially harmful treatment trials.the treating physician can avoid the use of drugs that are potential mitochondrial toxins and handle metabolic crisis situations diligently.

Although drug treatment is largely supportive and symptomatic,it can provide meaningful clinical benefit to patients . Our patient was started on a regimen of coenzyme Q10,creatine,carnitine,omega 3 fatty acid,vitamin E and folic acid. Co-Q10 transports electrons between complex I and complex III of the mitochondrial respiratory chain and has been shown to improve mitochondrial function in several studies [8]. Creatine generates additional ATP through the creatine phosphate shuttle. Carnitine enhances transport of fatty acids into the mitochondria. Folic acid is a cofactor for several mitochondrial enzymes (9). There was tremendous improvement in the symptoms of the patient,during the first few months.With continued therapy,patient had a gradual resolution of the disease symptoms.

CONCLUSION

This case postulates that mitochondrial disorder should be considered in a patient with recurrent episode of metabolic acidosis,after evaluating the other etiologies.

REFERENCES

1) Jung, B., Martinez, M., Claessens, Y. *et al.* Diagnosis and management of metabolic acidosis: guidelines from a French expert panel. *Ann. Intensive Care* 9, 92 (2019).
2) Andersen LW, Mackenhauer J, Roberts JC, Berg KM, Cocchi MN, Donnino MW. Etiology and therapeutic approach to elevated lactate levels. *Mayo Clin Proc*. 2013;88(10):1127-1140.
3) Rajasurya V, Anjum H, Surani S. Metformin Use and Metformin-associated Lactic Acidosis in Intensive Care Unit Patients with Diabetes. *Cureus*. 2019;11(5):e4739.
4) Chinnery PF, Johnson MA, Wardell TM, et al. The epidemiology of pathogenic mitochondrial DNA mutations. Ann Neurol. 2000;48(2):188-193.
5) Schaefer AM, McFarland R, Blakely EL, et al. Prevalence of mitochondrial DNA disease in adults. *Ann Neurol*. 2008;63(1):35-39.
6) Elliott HR, Samuels DC, Eden JA, Relton CL, Chinnery PF. Pathogenic mitochondrial DNA mutations are common in the general population. *Am J Hum Genet*. 2008;83(2):254-260.
7) Castro-Gago M, Alonso A, Pintos-Martínez E, et al. Lipomatosis simétrica múltiple asociada a polineuropatía, atrofia de cerebelo y citopatía mitocondrial [Multiple symmetric lipomatosis associated to polyneuropathology, atrophy of the cerebellum and mitochondrial cytopathy]. *Rev Neurol*. 2003;36(11):1026-1029.
8) Haas RH. The evidence basis for coenzyme Q therapy in oxidative phosphorylation disease. *Mitochondrion*. 2007;7 Suppl:S136-S145.
9) Tarnopolsky MA. The mitochondrial cocktail: rationale for combined nutraceutical therapy in mitochondrial cytopathies. *Adv Drug Deliv Rev*. 2008;60(13-14):1561-1567.