P-15 A case of adult onset Leigh syndrome presented with acute renal failure and severe metabolic acidosis

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Introduction

Generally, Mitochondria disease caused by mutations in mitochondrial DNA or nuclear DNA, is typically seen in infancy or childhood. In this case, we report a rare cases of adult onset mitochondria disease with acute renal failure.

Case Report

A 43-year-old man referred to our institution originally presented with loss of consciousness and disorientation. The family history showed no nervous system or muscle dysfunction. He was noted to have deteriorated physically, reporting shortness of breath during and after exertion. An arterial blood gas analysis showed the following: pH 7.268, 14.9 Torr PCO2, 98.3 Torr PO2, 6.7 mmol/L HCO3. Urinalysis showed protein-positive (3+) 16 g/g･Cr. His serum laboratory values were BUN 146 mg/dl, Cr 12.0 mg/dl, Na 130mEq/L, K 6.7mEq/L, and lactate 28.4 mg/dl. Noncontrast CT findings were almost normal. We used continuous hemodiafiltration for acute renal failure and metabolic acidosis. But his metabolic acidosis was sustained. He suddenly presented myoclonic seizures at 14 days and showed a low-density signal of left temporal lobe cortical lesion in diffusion-weighted (DW) magnetic resonance imaging (MRI) at 43 days. Moreover, he presented left eye deviation. The brainstem showed symmetrical hyperintense lesions in DW MRI at 56 days. Mitochondrial DNA analysis from a serum specimen showed G13513A mutation. Therefor we diagnosed LS and L-arginine treatment was started. But he had complete AV block at 63 days and led to dead at 73 days.

Conclusion

In this case, mitochondria disease caused by G13513A mutation with acute renal failure before the neurological episodes were a rare case. In cases of renal failure with refractory metabolic acidosis, recurrent convulsion, and multiple organ failure, we should suspect mitochondria disease.