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# Clinical Manifestations, Pathogenesis and Medical Treatment of Stroke Caused by Basic Mitochondrial Abnormalities (Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like Episodes, MELAS)

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**Abstract :** Aim This case aims to discuss the pathogenesis, clinical manifestations and medical treatment of strokes caused by mitochondrial gene mutations. Methods Diagnosis of ischemic stroke caused by mitochondrial gene defect by means of "next-generation sequencing mitochondrial DNA gene variation detection", imaging examination, neurological examination, and medical history; this study took samples from the neurology ward of a medical center in northern Taiwan cases diagnosed with acute cerebral infarction as the research objects. Result This case is a 49-year-old married woman with a rare disease, mitochondrial gene mutation inducing ischemic stroke. She has severe hearing impairment and needs to use hearing aids, and has a history of diabetes. During the patient’s hospitalization, the blood test showed that serum Lactate: 7.72 mmol/L, Lactate (CSF) 5.9 mmol/L. Through the collection of relevant medical history, neurological evaluation showed changes in consciousness and cognition, slow response in language expression, and brain magnetic resonance imaging examination showed subacute bilateral temporal lobe infarction, which was an atypical type of stroke. The lineage DNA gene has m.3243A>G known pathogenic mutation point, and its heteroplasmic level is 24.6%. This pathogenic point is located in MITOMAP and recorded as Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like episodes (MELAS) , Leigh Syndrome and other disease-related pathogenic loci, this mutation is located in ClinVar and recorded as Pathogenic (dbSNP: rs199474657), so it is diagnosed as a case of stroke caused by a rare disease mitochondrial gene mutation. After medical treatment, there was no more seizure during hospitalization. After interventional rehabilitation, the patient's limb weakness, poor language function, and cognitive impairment have all improved significantly. Conclusion Mitochondrial disorders can also be associated with abnormalities in psychological, neurological, cerebral cortical function, and autonomic functions, as well as problems with internal medical diseases. Therefore, the differential diagnoses cover a wide range and are not easy to be diagnosed. After neurological evaluation, medical history collection, imaging and rare disease serological examination, atypical ischemic stroke caused by rare mitochondrial gene mutation was diagnosed. We hope that through this case, the diagnosis of rare disease mitochondrial gene variation leading to cerebral infarction will be more familiar to clinical medical staff, and this case report may help to improve the clinical diagnosis and treatment for patients with similar clinical symptoms in the future.

**Keywords :** acute stroke, MELAS, lactic acidosis, mitochondrial disorders
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