

CLINICAL PHENOTYPES, LONG TERM FOLLOW-UP AND PROGNOSIS IN TAIWANESE PATIENTS WITH MITOCHONDRIAL DNA A3243G MUTATION

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Mitochondriopathies are multi-system disorders and are related to either sporadic, mitochondrial DNA or nuclear DNA mutations. The clinical manifestations of mitochondrial disease are variable but some mitochondrial syndromes have been well recognized. Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) is the most common mitochondrial syndrome in Chinese patients and is usually associated with A3243G mutation. In the past decades, the pathogenesis, clinical features, image findings, and diagnostic tools have been broadly studied. However, only few clinical reports reviewed the prognosis of the MELAS patients with A3243G mutation especially in Taiwanese patients. A previous review concluded that the prognosis of A3243G mutation patients was correlated to the concentration of mutant mtDNA and the prognosis was worse in patients with status epilepticus. We performed a longitudinal study in 25 patients with A3243G mutation. Some developed complete or partial MELAS syndrome but others presented with hearing impairment or diabetes only. The prognosis remains poor if stroke-like episodes or seizure attacks occurred frequently.

Keywords:

MELAS, A3243G, mitochondrial disease, prognosis, Taiwanese