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Whole mitochondrial DNA sequencing of 47 patients with Leigh syndrome without prevalent 8993T>G, 8993T>C, 9176 T>C or 13513G>A mutations

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8993T>G, 8993T>C, 9176 T>C, 13513G>A mutationsを認めないLeigh syndrome 47例におけるミトコンドリア全周シークエンス解析

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Leigh syndrome (LS) is a neurodegenerative disorder in infancy or childhood characterized by focal, bilaterally symmetric necrotizing lesion of basal ganglia, thalamus and brainstem. LS is caused by mutations in mitochondrial or nuclear genes involved in mitochondrial energy metabolism. To investigate the significance of mitochondrial DNA mutations in LS, we performed whole mitochondrial DNA sequence analysis in 47 patients with LS who had no 8993T>G, 8993T>C, 9176 T>C or 13513G>A mutation relatively prevalent in LS (approximately 20%). The age is between 4 months to 65 years old (median 2 years old), 21 males and 26 females. We identified 5 mutations previously reported as a cause of LS in 6 patients (8363G>A, 10158T>C, 10197C>A, 11777C>A each in one patient; 14459C>A in 2 patients). In addition, we detected other 4 mutations which have been neither reported as a cause of disease nor a polymorphism, and it is necessary to analyze the mutation effects on mitochondrial functions to judge the pathogenesity of the mutations. Whole mitochondrial DNA sequencing is useful for diagnosis of LS caused by rare mitochondrial DNA mutation and also important information for genetic counseling.

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