Large-scale mitochondrial DNA deletions in patients with CPEO syndrome in taiwan.

謝榮鴻
Wang EK;Kao KP;Hsieh RH;Lu CY;Pang CY;Wei YH.

Abstract

We investigated the mtDNA mutations in muscle biopsies obtained from eight Chinese patients with chronic progressive external ophthalmoplegia by Southern blot analysis and direct DNA sequencing. The results showed that a common 4,977 bp deletion is present in the muscle of ve of these patients and the proportions of the mutant mtDNA were 76%, 46%, 66%, 60%, and 86%, respectively. The patients who harbored higher proportions of the 4,977 bp deleted mtDNA tended to have more severe clinical phenotype and earlier onset of the disease. This is the rst report of Chinese patients with CPEO syndrome who are associated with the 4,977 bp deletion of mtDNA. Moreover, these ndings are consistent with the previous reports that large-scale deletions of mtDNA are present in the target tissues of patients with CPEO syndrome.