DELETIONS OF MITOCHONDRIAL DNA IN KEARNS-SAYRE SYNDROME

M. Zeviani, C. T. Moraes, S. DiMauro, H. Nakase, E. Bonilla, E. A. Schon, and L. P. Rowland

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We have identified large-scale deletions in muscle mitochondrial DNA (mtDNA) in seven of seven patients with Kearns-Sayre syndrome (KSS). We found no detectable deletions in the mtDNA of ten non-KSS patients with other mitochondrial myopathies or encephalomyopathies, or three normal controls. The deletions ranged in size from 2.0 to 7.0 kb, and did not localize to any single region of the mitochondrial genome. The proportion of mutated genomes in each KSS patient ranged from 45% to 75% of total mtDNA. There was no correlation between the size or site of the deletion, biochemical abnormality of mitochondrial enzymes, or clinical severity. The data bolster arguments that KSS is a unique disorder and genetic in origin.

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Comment from Andrew G. Engel, MD, FAAN, Associate Editor: This article set the stage for the diagnosis of mitochondrial myopathies due to large-scale deletions in mitochondrial DNA.

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