Mitochondrial complex I – linked disease

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Mitochondrial complex I – linked disease

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Abstract

Complex I deficiency is the most frequently encountered single mitochondrial single enzyme deficiency in patients with a mitochondrial disorder. Although specific genotype-phenotype correlations are very difficult to identify, the majority of patients present with symptoms caused by leukodystrophy. The poor genotype-phenotype correlations can make establishing a diagnosis a challenge. The classical way to establish a complex I deficiency in patients is by performing spectrophotometric measurements of the enzyme in a muscle biopsy or other patient-derived material (liver or heart biopsy, cultured skin fibroblasts). Complex I is encoded by both the mtDNA and nuclear DNA and pathogenic mutations have been identified in the majority of the 44 genes encoding the structural subunits of complex I. In recent years, the increasing possibilities for diagnostic molecular genetic tests of large gene panels, exomes, and even entire genomes has led to