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Diagnosis of mitochondrial disorders applying massive pyrosequencing.

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Abstract

Mitochondrial disorders are a frequent cause of neurological disability affecting children and adults. Traditionally, molecular diagnosis of mitochondrial diseases was mostly accomplished by the use of Sanger sequencing and PCR-RFLP. However, there are particular drawbacks associated with the use of these methods. Recent multidisciplinary advances have led to new sequencing methods that may overcome these limitations. Our goal was to explore the use of a next generation sequencing platform in the molecular diagnosis of mitochondrial diseases reporting our findings in adult patients that present with a clinical-pathological diagnosis of a mitochondrial encephalomyopathy. Complete genomic sequences of mitochondrial DNA were obtained by 454 massive pyrosequencing from blood samples. The analysis of these sequences allowed us to identify two diagnostic pathogenic mutations and 74 homoplasmic polymorphisms, useful for obtaining high-resolution mitochondrial haplogroups. In summary, molecular diagnosis of mitochondrial disorders could be efficiently done from readily accessible samples, such as blood, with the use of a new sequencing platform.

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