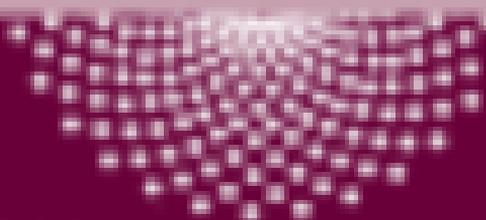


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The Relation of Mitochondrial DNA Mutation with Mitochondrial Diseases in Coding Region

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Abstract

Mitochondrial disorders are recognized in several metabolic and degenerative diseases, aging and cancer. Mitochondrial functional deficiency maybe caused by a decrease in the function of complex respiratory enzymes that can inhibit the oxidative phosphorylation chain (OXPHOS) for synthesis of ATP. Based on data MITOMAP in 2013, there are 211 of 466 mutations that have been reported in coding region. The largest number of mutation located on MT-COI region that have 34 mutations. In particular, mutations in a subunit of COX have been described associate with various clinical phenotypes. Deficiency of COX is one of disorder that often leads to mitochondrial disease. Based on the database, the dominant disease on subunit of COI is dominated by prostate cancer. Moreover, the highest probability of mutation to the size of gene located on MT-NDI with 2.83%. OXPHOS deficiency which is caused by DNA mitochondrial mutation, mostly appears on subunit NDI. The dominant phenotype on subunit of NDI is Leber's hereditary optic neuropathy (LHON) diseases. This study has revealed that mutations in mitochondrial DNA were not only associated with predisposition to neuromuscular diseases but also to cancer and optical interference.

Keywords: Mitochondrial DNA, Mitochondrial disease, subunit of COI, Prostate Cancer, subunit of NDI, LHON

1. Introduction

Mitochondria is a sub cellular organelle which is responsible over mostly adenosine triphosphate (ATP) synthesis in cell through oxidative phosphorylation (OXPHOS)¹. Moreover, mitochondria is an important biochemistry pathway, including tricarboxylic acid cycle (TCA) and urea cycle. Mitochondria is also crucial as a regulator of apoptosis process², calcium cytosolic concentration and the central biogenesis of iron-sulphur cluster (Fe-S)¹. Mitochondria disorder is known as the most significant contribution of metabolism and degenerative diseases, aging and cancer^{3,4,5}.

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