

## PROFIL GEN ATPase 6 DNA MITOKONDRIA MANUSIA PADA POPULASI DATARAN TINGGI

### PROFILE OF MTDNA ATPase 6 GENE IN HUMAN POPULATION OF HIGH ALTITUDE

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#### ABSTRACT

*Studies on the variability mitochondrial DNA (mtDNA) genes are usually focused to answer the question of whether selection plays the dominant role to shape the genetic profile. The detection of ATPase 6 gene indicate that variations in genes do not necessary causing negative effect to individual, instead it contributes to the adaptation process in the individual's habitat. This study was aimed to find characteristic profile of mtDNA ATPase 6 gene in human population of high altitude. Result showed different pattern of sequence and amino acid variations of ATPase 6 gene between populations from the same subgroup but living in different altitude. Based on nucleotide sequence analysis, it showed higher number of variation (mutation number, position and type) of the ATPase 6 gene in population of higher altitude compared to those in lower altitude. There were six point mutations potential characteristic variation, three of them A8701G, A8730G and A8860G were demonstrated in higher frequency, which made them candidates of characteristic ATPase 6 gene profile of human population in high altitude.*

**Key words:** mtDNA, ATPase 6 gene, high altitude, polymorphism

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