

## SUMMARY

### KETERKAITAN POLIMORFISME GEN ATP SINTASE 6 DENGAN KETINGGIAN GEOGRAFIS

Heli Siti H.M.\*, Yoni Fuadah Syukriani\*\*, A.S. Noer\*\*\*, Gun Gun Gumilar\*

\*Program Studi Kimia, FPMIPA UPI

Jl. Dr. Setiabudhi 229-Bandung

[heli@upi.edu](mailto:heli@upi.edu)

\*\*Fakultas Kedokteran Forensik UNPAD

\*\*\*Departemen Kimia FMIPA ITB

\*\*\*\*Departemen Biologi SITH ITB

Studies on the variability mtDNA genes are usually focused to answer the question of whether selection plays the dominant role to shape the genetic profile. The detection of ATPase6 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. The ATPase6 variants have been understood to affect the increase of basal metabolism, particularly in the arctic populations. Nevertheless, there is still not enough study that is trying to find the relation between hypoxic habitat with mtDNA genetic variants, particularly within the ATPase6 gene; even though the essential role of oxygenation in mitochondrial energy metabolism through OXPHOS is well understood. This study was aimed to find characteristic profile of mtDNA ATPase6 gene in human population of high altitude. Characteristic gene profile will indicate ATPase6 gene's relationship with individual resistance to oxygen level; the knowledge has a potential to contribute on re-engineering of therapy or substance that may support better oxygen metabolism. Result showed different pattern of sequence and amino acid variations of ATPase6 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 ATPase6 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 ATPase6 gene profile of human population in high altitude. This first year findings suggest the relationship between variations of ATPase6 gene and high altitude, which showed the need to find the molecular mechanism of ATPase structural changes through protein analysis of the variations and confirm it by using other variations from different high altitude populations. The finding of an ATPase6 amino acid change from threonin to alanin suggested that modelling of F0 ATPase6 3D structure, which was planned as one of the second year activities,

is necessary to answer the question of whether adaptation to high altitude plays a role in OXPHOS function.