

# HGM 2015 - Abstract Submission

*Topic: Populations Genetics*

HGM15ABS-1065

## **ASSOCIATION OF SINGLE NUCLEOTIDE POLYMORPHISMS (SNP) IN PCSK9 GENE WITH LIPID ANALYSIS AMONG IBAN AND BIDAYUH ETHNIC GROUPS IN SARAWAK POPULATION**

H. H. Hood\*<sup>1</sup>, Y. T. Siaw<sup>1</sup>, S. P. Sim<sup>1</sup>, H. Helmy<sup>2</sup>, M. M. Aminudin<sup>3</sup>

<sup>1</sup>Paraclinical Sciences, Universiti Malaysia Sarawak, Kuching, <sup>2</sup>Community Medicine and Public Health, <sup>3</sup>Centre for Pre-University Studies, Universiti Malaysia Sarawak, Kota Samarahan, Malaysia

**Preferred presentation method:** Oral or poster

**Would you like to apply for a travel grant?:** Yes

**Objectives:** Proprotein convertase subtilisin/kexin type 9 (PCSK9) gene (MIM: 603776) encoded protein that facilitates cholesterol level in blood. Known mutation in the PCSK9 gene would indicate hypercholesterolemia through gain of function resulting in Familial Hypercholesterolemia Type 3. The increased risk genetic variant: rs 12084215 of FH (Say et al, 2013) were used to indicate any correlation with performed lipid analysis with untested indigenous population in Sarawak. We are conducting a study to determine the polymorphic genotype frequencies among 2 indigenous populations, the Iban and the Bidayuh in Sarawak, thus elucidating any association with lipid analysis.

**Methods:** Blood samples from study subjects were collected for both genotypes and lipid profile. Peripheral blood samples were collected, genomic DNA extracted and genotyped employing Allele Specific-PCR. Genotypes were categorized into homozygous wild type, heterozygous and homozygous variants. The lipid profile consists of total cholesterol, high density lipoprotein, low density lipoprotein, and triglycerides. Statistical analyses were done to determine the genotype frequencies and association with polymorphic genotypes with lipid analysis.

**Results:** Total of 112 candidates were assessed to be 92 (82.1%) homozygous wildtype and 20 (17.9%) heterozygous SNP. There were no significant association between the genotypes and lipid profiles. The intronic genetic variant may not affected the gene expression thus explained why the lipid profiles of tested population lies within the optimal classification provided by ATP III guidelines. The guidelines give information for global risk assessment in coronary heart diseases (CHD) and other lipid related disorders.

**Conclusion:** Study present limited information to associate the polymorphic genotype frequencies with lipid profiles. Thus, more samples and SNPs in PCSK9 genes as well as other lipid related gene are required to provide sufficient data for the analysis. Besides that, more works should be done to elucidate the genes responsible for lipid abnormality.

**Disclosure of Interest:** None Declared