

HGM 2015 - Abstract Submission

Topic: Populations Genetics

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SINGLE NUCLEOTIDE POLYMORPHISMS (SNPS) OF LOW-DENSITY LIPOPROTEIN RECEPTOR GENE (LDLR) IN LIPID RELATED GENE-ASSOCIATED FAMILIAL HYPERCHOLESTEROLAEMIA AMONG IBAN AND BIDAYUH ETHNIC GROUPS IN SARAWAKIAN POPULATION: PRELIMINARY DATA ANALYSIS

Y.T Siaw^{*1}, H.H Hood¹, S. P Sim¹, H. Helmy², M. M. Aminudin³

¹Paraclinical Science, ²Community Medicine and Public Health, ³Centre for Pre-University Studies, Universiti Malaysia Sarawak, Kuching, Malaysia

Preferred presentation method: Oral or poster

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

Objectives: Familial Hypercholesterolaemia (FH) is a genetic disease caused by defects in a number of variants, including low-density lipoprotein receptor gene (*LDLR*) that regulate plasma LDL-cholesterol concentrations. FH typically passed down through families in an autosomal dominant manner. That means one only needs to acquire the abnormal gene from one parent in order to be affected by the disease. FH has a worldwide prevalence of 0.2%, however it is considerably higher in some population because of a founder effect. This study was initiated as there is no FH data available in Sarawak. We aimed to determine the frequencies lipid related gene polymorphisms in Iban and Bidayuh ethnic groups in Sarawak, and its associations with lipid profiles.

Methods: A total of 110 Iban and Bidayuh were recruited. Physical assessments were performed and two blood tubes were withdrawn. Subjects' clinical blood lipid parameters were analyzed and DNA was extracted. Allele Specific-PCR of *LDLR* gene for two polymorphisms (c. 1060+7 T>C & c. 1706-55A>C) were categorized into homozygous wild, heterozygous and homozygous SNP. Agarose gel electrophoresis was performed and the data were analysed using SPSS.

Results: Preliminary data shows that 99.1% have homozygous SNP and 0.9% have homozygous wild in c. 1060+7 T>C, while in c. 1706-55A>C: 66.4% have heterozygous SNP, 21.8% have homozygous SNP and 11.8% have homozygous wild. So far there is no significant association between SNPs and lipid profiles level among Iban and Bidayuh population in Sarawak. We also found out that the level of LDL mean is approximately and within the border line for all the categories of genotypes. The mean (SD) of LDL cholesterol level for c. 1060+7 T>C homozygous wild, homozygous SNP is 3.4 (0.00) and 3.2 (1.09) respectively while for c. 1706-55A>C heterozygous SNP, homozygous wild and homozygous SNP is 3.2 (1.05), 3.4 (1.03) and 3.3 (1.25) respectively. This might be due to the combined genotypes of variants within *LDLR* gene were detected.

Conclusion: Preliminary findings from this ongoing research show that SNPs of the *LDLR* gene are present in the Iban and Bidayuh. Further works is needed to determine the linkage disequilibrium among variants of *LDLR* gene.

Disclosure of Interest: None Declared