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**Analysis of Genetic Variants in a Malay Family With Hereditary Nonpolyposis Colorectal Cancer (HNPCC)**

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**ABSTRACT**

Hereditary nonpolyposis colorectal cancer syndrome (HNPCC), is an inherited tendency to develop colorectal, endometrial (uterine) and other cancers. Although most cancers are not inherited, about 5 percent (%) of individuals with colorectal or endometrial cancers have HNPCC. HNPCC is caused by alteration in the mismatch repair (MMR) genes; MLH1, MSH2, MSH6 and PMS2. To date, there is no published study on HNPCC familial mutations specifically on the Malay population, the biggest ethnic group in Malaysia. Therefore, this study aimed to identify the genetic variants in a Malay HNPCC family. Two probands enrolled into this study fulfilled at least one of the Bethesda criteria Both probands were first degree relative diagnosed with colorectal cancer. The whole genome sequences of these probands were sequenced by using Illumina HiSeqX. Single nucleotide variants (SNVs) were defined and characterised. Ten genetic variants were identified in both probands with three genetic variants found in MSH2 gene; two genetic variants found in MSH6 gene and five genetic variants identified in PMS2 gene. Among all the genetic variants identified in this family, only one variant in the MSH6 gene; c.116G>A (p.Gly39Glu) which is located in exon 1, was found in both probands. The genetic variant has been previously reported to be associated with an increased risk of colorectal cancer in male patients (Campbell et al. 2009). However, as yet, no study on Malay cohorts have been reported. This present study have identified a particular genetic variant in the MSH6 gene; c.116G>A (p.Gly39Glu), which has yet to be reported in any Malaysian colorectal cancer cases. In addition, our results also provide additional knowledge on genomic spectrum that highlights the hereditary predisposition of colorectal cancer.