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**The Landscape of Somatic Mutations in Malaysian Colorectal Cancer Patients using Whole Genome Sequencing**

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**ABSTRACT**

Over the past few years, colorectal cancer (CRC) remains as the third most common cancer worldwide and the incidence is continuously increasing in many countries including Malaysia. The rise in CRC burden in Malaysia particularly, underlines the importance of pushing ahead in our pursuit of understanding this cancer by profiling the genome of local CRC patients. Many of the published data on whole genome sequencing of CRC are not from this side of the world. In this study, we aimed to characterize the landscape of somatic mutations using whole genome sequencing approach and to identify the key molecular networks involved using our local CRC patients’ samples. Whole genome sequencing of at least 30X coverage, was performed on genomic DNA obtained from 13 pairs on matched tumour and corresponding blood samples. We detected all types of somatic mutations including non-silent single nucleotide variants (SNVs), insertions and deletions in both coding as well as in non-coding regions of the genome. We extended the mutational signature analysis to genomic rearrangement. We obtained an average of 29 indels, 318 SNVs and 78 genome rearrangements in tumor samples. We discovered that TP53 (10/13), APC (9/13) and KRAS (5/13) appeared as the most commonly mutated genes in our cohort of patients. Wnt signaling pathway is the major affected pathway. Interestingly, we identified recurrent 14bp frameshift deletions in the FGFR3 gene (p551fs) in six patients and 10bp frameshift insertions in FGFR1 gene (M578fs) in two patients. All variants identified in both FGFR1 and FGFR3 were located at the tyrosine kinase domain of the genes. This analysis of mutational signatures from our own local patients illustrates a multidimensional and comprehensive genomic landscape that highlights the molecular complexity of colorectal cancer and provides a road map to facilitate genome guided personalized therapy in Malaysian population.