



UNIVERSITI
TEKNOLOGI
MARA

THE DOCTORAL RESEARCH ABSTRACTS

Volume: 11, Issue 11

April 2017

ELEVENTH ISSUE

INSTITUTE of GRADUATE STUDIES

IGS Biannual Publication



Name : ROSE ISZATI BINTI ISMET NAYAN

Title : WHOLE GENOME SEQUENCING AND ANALYSIS ON THE TRIOS OF THE CHE WONG AND SEMAI: UNDERSTANDING THE MEDICO-GENOMIC ASSOCIATION

Supervisor : PROF. DATO' DR. MOHD ZAKI SALLEH (**MS**)
PROF. DR. TEH LAY KEK (**CS**)

As to date, there is no report on the architecture of the whole genomes of the unique Orang Asli subtribes in Peninsular Malaysia. The Che Wong was selected as the group of interest as they have a dwindling population of 651, while the Semai Orang Asli with a population number of 51,313 was selected as the control group. The Semai is the largest sub-tribe of the Senoi which is also the largest group amongst the Orang Asli. This study aims to understand the variation in genomics composition of the Che Wong and Semai Orang Asli by a deep and systematic characterization of the genome through development of an in-house bioinformatics pipeline. The disease risk and protection conferred by genetic traits were explored via sequencing the whole genome of a trio family of both the Che Wong and Semai subtribes. The Che Wong and Semai genomes were sequenced and mapped to the human reference genome (hg19) with an average coverage of 44.1x and 43.0x, respectively. A total of ~6.23 million and ~6.18 million variants were identified for the Che Wong and Semai trios, respectively. An average of 448,166 and 436,520 of these variants were found to be unique to the

Che Wong and Semai trios, respectively. An average of 1.6% of the total variants called for the Che Wong and Semai genomes were known to be evolutionary conserved. A closer relationship between the Che Wong and Semai with the Asian populations than the African, American and European peoples was also observed. There is a distinctive difference in health status between the Che Wong and Semai trio where the Semai are seen to be in a healthier state. Medico-genomic association also revealed more disease impact for the Che Wong trio as compared to the Semai trio genomes. A total of 93 and 106 *de novo* mutations (DNMs) were identified for the Che Wong and Semai trio respectively. These mutations were also studied for their medico-genomic association. We report here for the first time the catalogues of the genomic architectures of the Orang Asli, the Che Wong and Semai in precise along with their medico-genomic findings. These data provide new perspectives of the genomics background of the indigenous populations in South East Asia (SEA) which we believe would be useful for the scientific and health community.